Welcome

NHS East Genomics

Making a Difference:
Genomics in Nursing and Midwifery Conference

Tuesday 22nd November 2022

Mercure Leicester, The Grand Hotel

Venue Wifi:

Network: Conference

Password: Mercure24

Twitter:

#EastGenomicsNurseMidwife







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NHS
Genomic Medicine Service

Nursing and Midwifery Genomic Transformation Programme

Professor Janice Sigsworth – Professiona Lead for Genomics in Nursing

Dr Naomi Chapman – Director of the National Nursing and Midwifery Genomic Transformation Programme

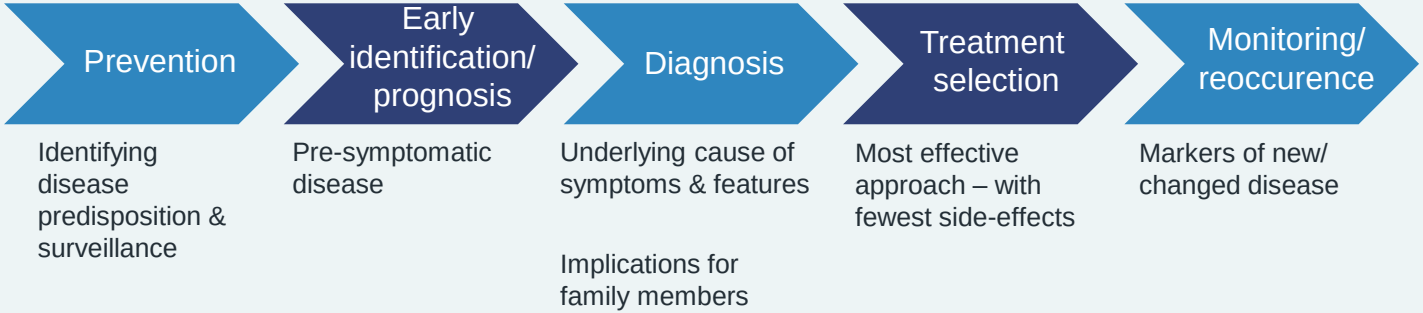
Why are we here?

This breakout session will give:

1. An overview of the organisations and structures in place to support genomics in the NHS
2. Outline the National Nursing and Midwifery Genomic Transformation Programme – how to get involved

What is genomics?

Genomics is providing a more detailed understanding of what causes illness and infectious disease and is underpinning the development of new interventions



Ellie-Rose

Young Brit Francesca Jones – who is missing two fingers and three toes because of a rare condition – storms into the main draw of the Australian Open, defying the doctors who told her to FORGET a tennis career

NHS

Mail Online

Lee

'Doctors saved my sight by injecting a gene into my eye': Experts say this cutting-edge technique could one day also help many with age-related vision loss

Aim of the National Nursing/Midwifery Programme

To integrate genomics into mainstream nursing and midwifery clinical practice in England's NHS

2021-2024

NHS England and NHS Improvement Genomics Unit sets out the strategic direction which supports delivery of the NHS GMS and commissions work to be delivered through GMSA infrastructure

Two broad areas:

1 National Nursing/Midwifery 'delivery programme' to transform clinical practice in specified pathways

2 Professional senior nursing/midwifery leadership, strategy, influence and support

National collaboration to integrate genomics in Nursing and Midwifery professions

‘Changes needed to accelerate integrating genomics across nursing and midwifery are complex.

Common themes lend themselves to a coordinated and collaborative strategic approach to sustained change’
Tonkin et al; 2020

Bringing together commissioning, delivery and the professional leadership in a collaborative partnership.

Each informs the other in a continuous cycle to ensure consensus, offering the best approach to accelerating the integration of genomics in practice

Chief Nurse Leads

Chris Morley

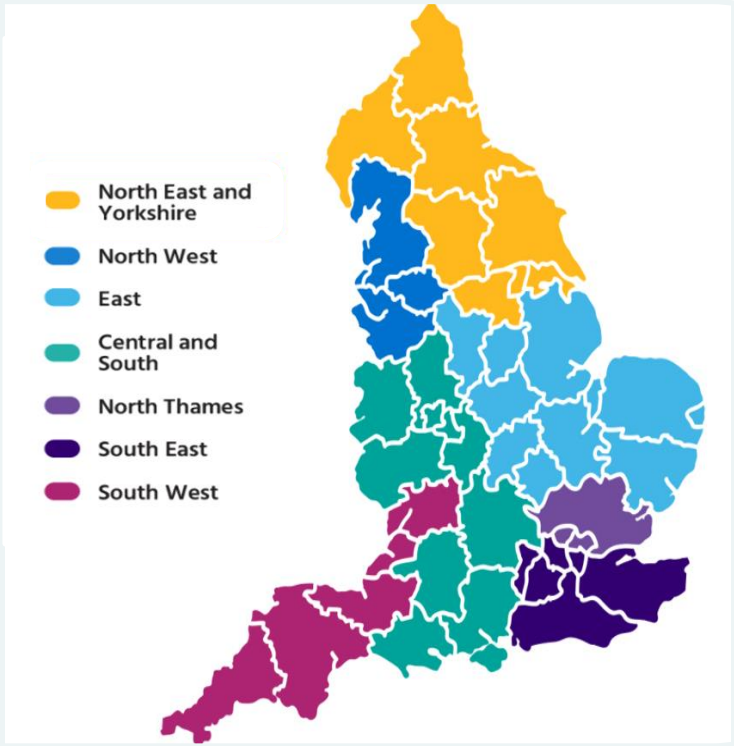
Janelle York

Lorraine Szeremeta

Lisa Stalley-Green

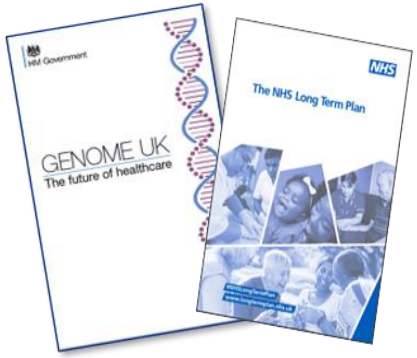
Avey Bhatia

Dr Carolyn Mills



Engaging senior leaders:

importance of strategic roles/influence in advocating change to integrate genomics



Key principles:

- Nurses and midwives leading change at every level and all settings
- Narrative and language - making it relevant to area of practice

- Align with CNO and CMidO strategic plans for change
- Visible leadership
- National professional lead – Professor Janice Sigsworth and Chief Nurses (one per GMS Alliance)

To support change we have:

- Developed new infrastructures – nurse & midwife leaders in new roles
- Designed the system to encourage collaboration, sharing, learning and networking

Strategic Leadership and National Infrastructure

CHIEF NURSES
in each GMS Alliance
(Chief Nurse on Alliance Partnership Board)

Chief Nurse Advisory Group
7 Chief Nurses in the group
Hosted by NHSE/I

A forum for delivery
(Hosted by NEY GMSA Delivery Team)
A group to support Lead GMSA Nurses as they deliver project outcomes/KPIs

OPERATIONAL LEAD NURSES/MIDWIFE
(in each Alliance)

PROJECT DELIVERY TEAM

Supports GMS Alliance teams and transfers methodology/ knowledge

Works with local teams in collaboration on projects to integrate genomics in selected priority clinical areas

National Nursing and Midwifery Genomic Transformation Programme

NHSE Genomics Unit (Chief Scientific Officer) has also commissioned a 2 year Nursing and Midwifery Transformation Programme.

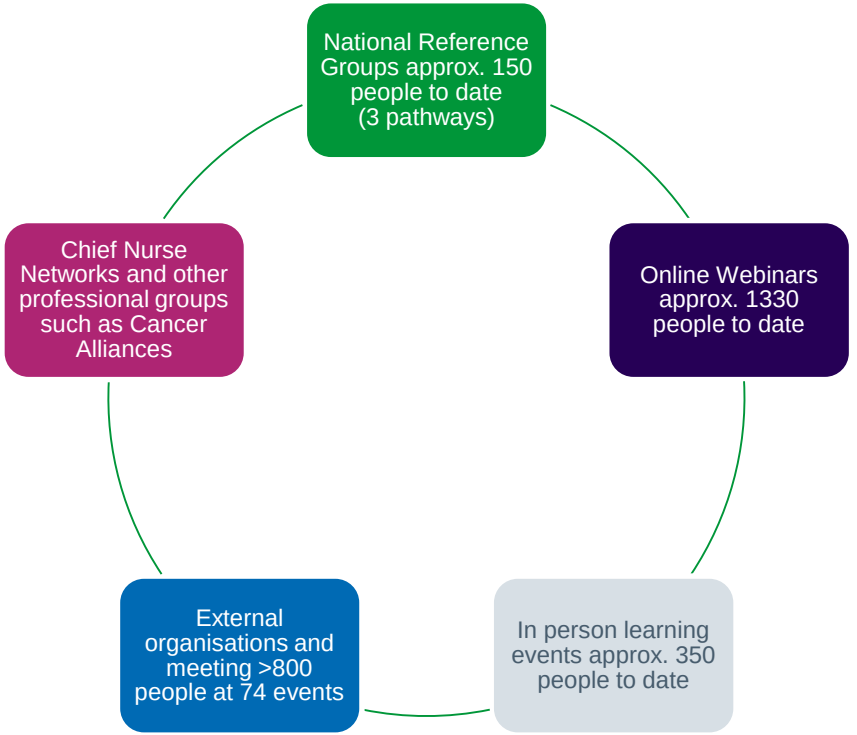
This programme is led by Dr Naomi Chapman and Dr Emma Tonkin

Aims –

- **Explore the nursing/midwifery inputs across 7 cancer or rare disease pathways with a range of outputs around pathway, education, patient perspective and the further contribution of nurses/midwives to the genomic conversations/ genomic testing**
- **Broaden genomic knowledge and skills within NHS mainstream care**

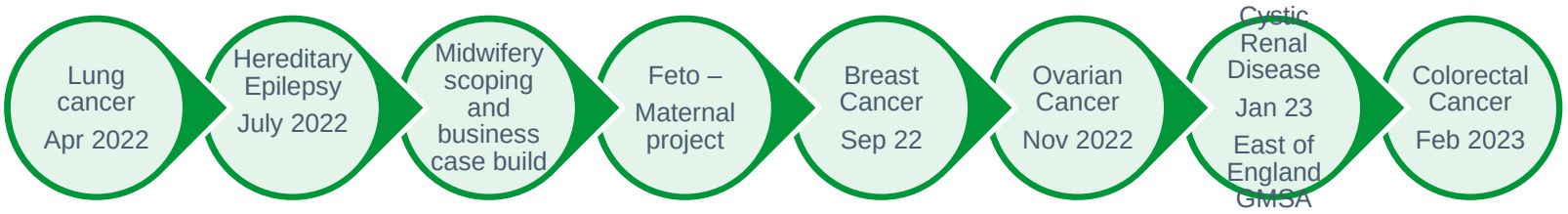
Engagement across England

- Working with Nurses and Midwives from all GMSAs to spread knowledge and input to programme to hundreds of participants.
- National Reference Groups formed for Pathways.
- Outputs shared for validation with key Nursing and Midwifery groups prior to wider sharing.
- Attendance at a wider range of key events and conferences to present or host stands with over events reaching over 1,830 colleagues
- Establishing and growing clinical networks.



Data for Webinars, In-person learning events and Meetings from 3 of 7 GMSAs

Pathway projects 2022-24



GMSAs bid for to lead one of the pathways.

First pathway began
Apr 2022 - Lung

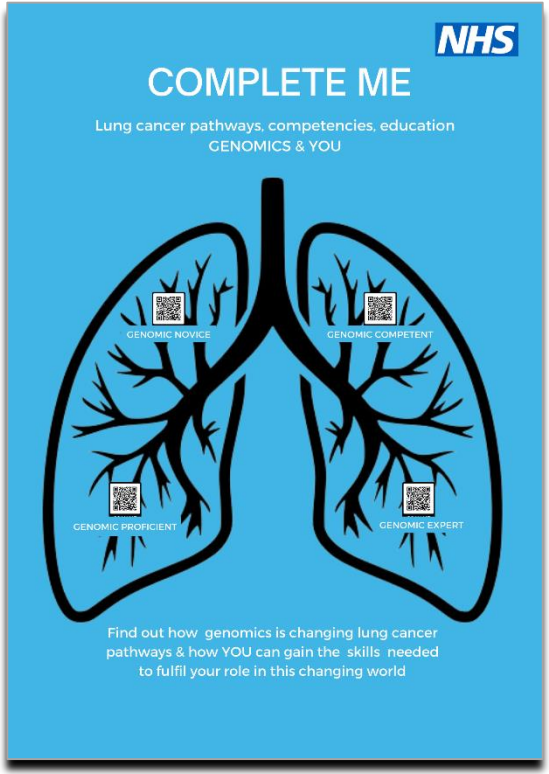
Milestones time line for each pathway defined and shared

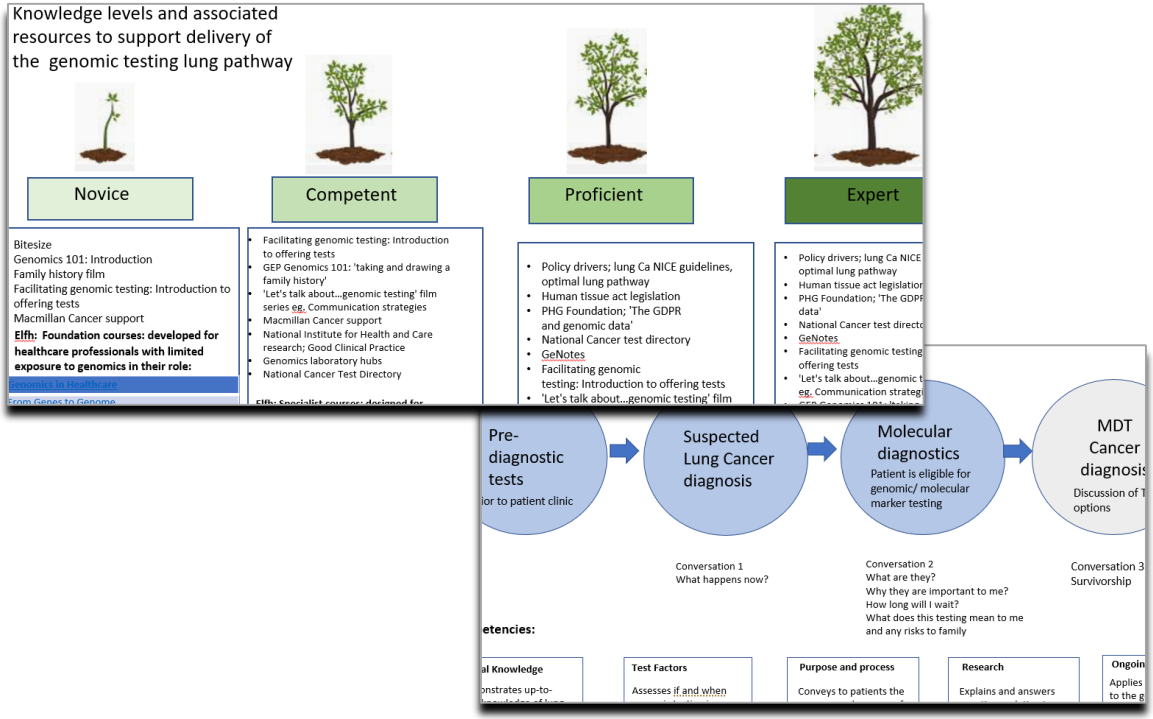
The Sprint – National Workshops

What will Pathways produce to support genomics in clinical care?

- Clinical Pathway Initiative - overview of key nursing/midwifery inputs. Mapping the education available to give the knowledge needed by those delivering the pathway.
- Produce a ‘patient story’ to illustrate the patient and family experience.
- Produce information to inform the GeNotes national programme of education for clinicians.
- Identify opportunities for development of genomic practice in care. Consider Equity of Access.
- Bring together a national reference group of clinicians in each pathway who form the membership of the workshops coming together to inform each pathway.

Pathways materials for dissemination





- Working collaboratively with GEP and HEE for Clinical Pathway Competencies Initiative and Educational Signposting.
- GENotes Production or Review

Thank you

Contacts

Vicky Carr

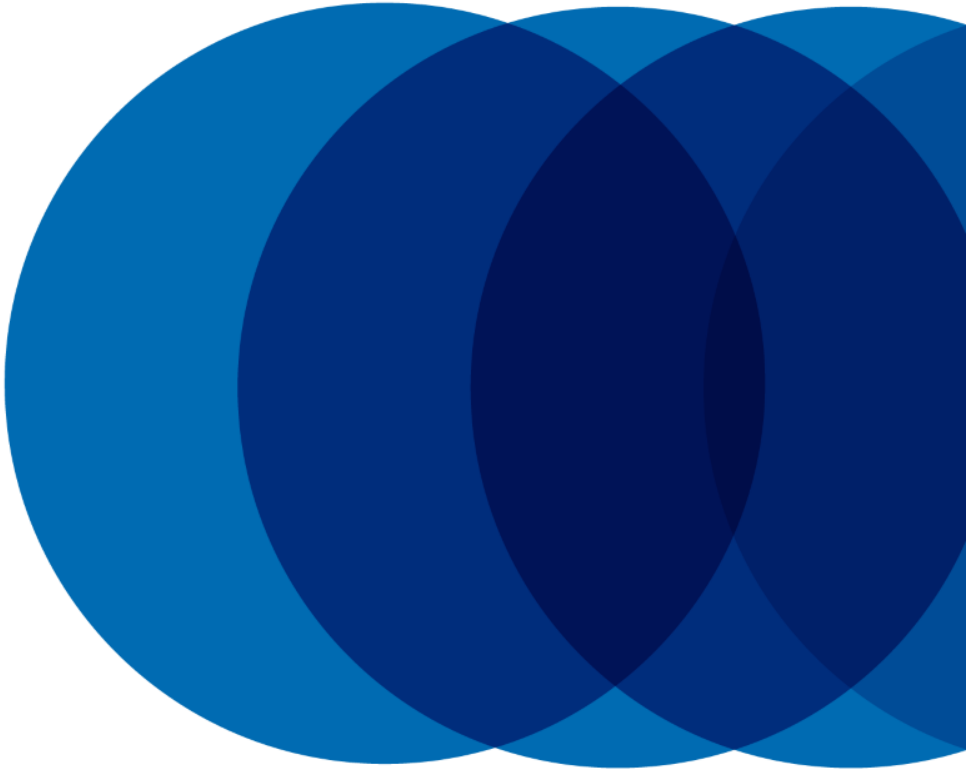
victoria.carr6@nhs.net

Dr Naomi Chapman

Naomi.chapman7@nhs.net

Dr Emma Tonkin

Emma.Tonkin@southwales.ac.uk



NHS
East Genomics

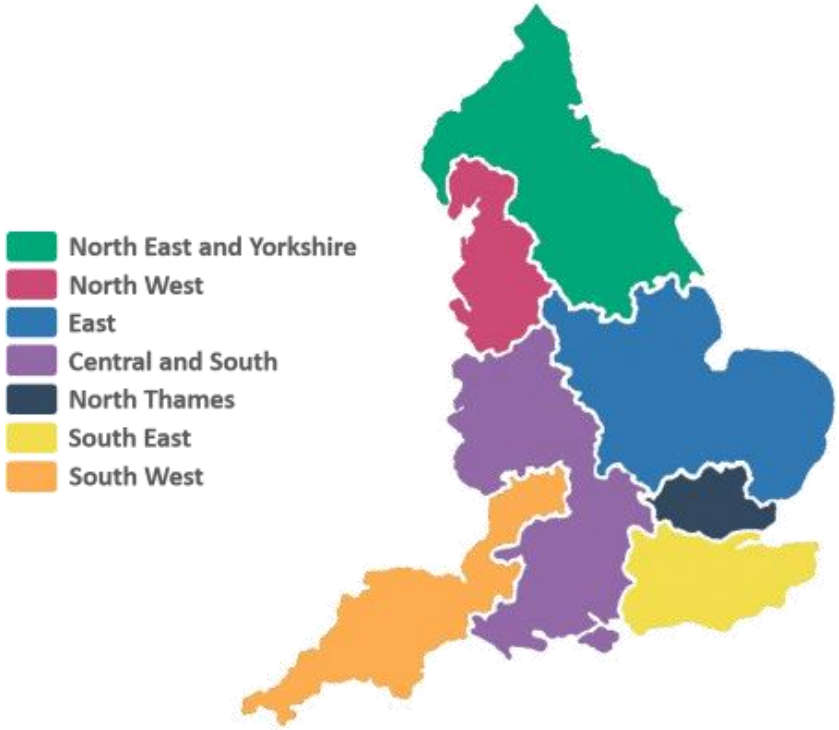
Annette Breen

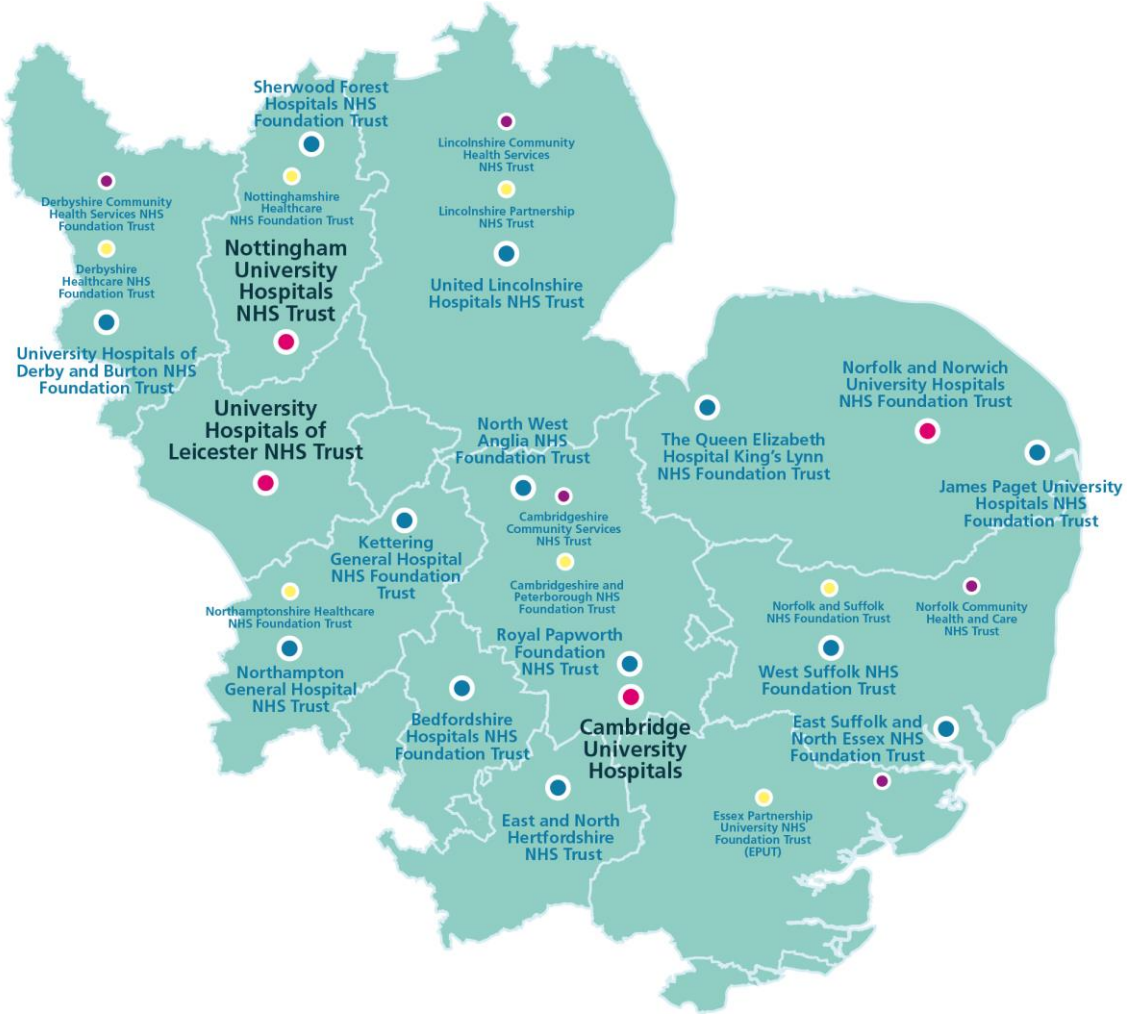
What is the East Genomic Medicine Service Alliance (GMSA)?
What is it doing?

Genomic Medicine Service Alliance (GMSA)

NHS
East Genomics

- Support the systematic embedding of genomics into routine clinical care
- Facilitate rapid adoption of scientific advances
- Collaborative partnership working across disciplines and geographies





NHS East Genomics

NHS East GMSA

●	4 partner orgs
●	Other acute trusts in the area *
●	Mental health trusts
●	Community trusts

* Includes Royal Papworth (specialist trust)

Genomic Medicine Services Alliances Aims

NHS East Genomics

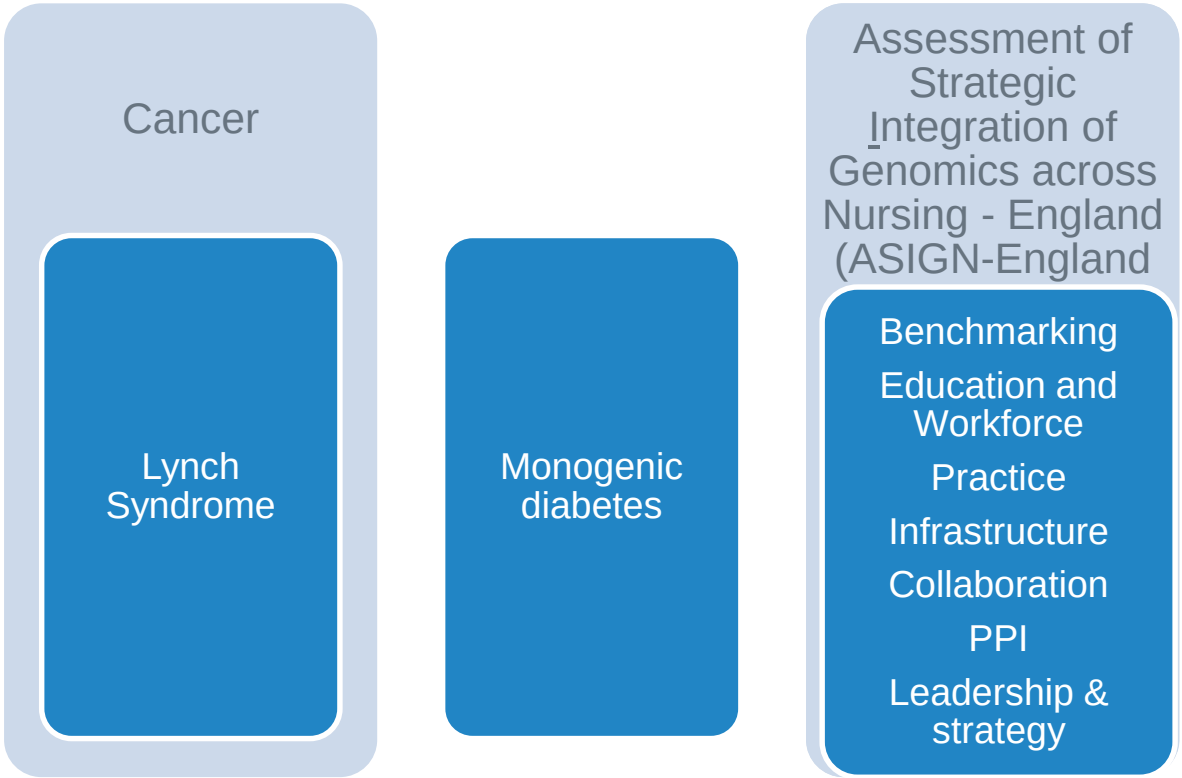
Their key aim is to achieve demonstrable improvements across the whole geography:

- Equitable access to standardised end-to end pathways
- Access to treatments and medicine optimisation driven by genomics
- Access to clinical trials by ensuring systematic consideration of eligibility
- Active participation and contribution to nationally co-ordinated with facilitated approach to genomic research

National Transformation Projects 2021-22

East Genomics

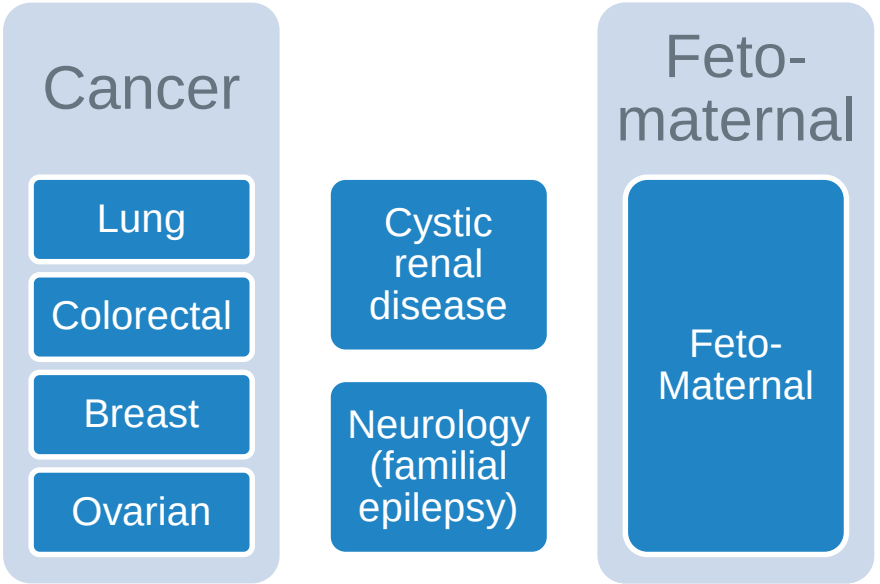
- Baselining, developing plans to mainstream genomics from highly specialist services to the wider healthcare team
- Increase equity of access and reduce unwarranted variation



National Transformation Projects 2022-24

East Genomics

- Delivery of change in practice at ‘key touchpoints’ to accelerate patient access to genomic testing where eligible and advantageous to inform predisposition to disease, treatments, prognosis, reproductive choices
- Increase equity of access and reduce unwarranted variation
- Inform best practice guidance for genomic pathways
- Identify learning and development needs and opportunities



Polycystic kidney disease (PKD)

East Genomics

- Autosomal dominant PKD leads to cysts in the kidneys and liver, and sometimes pancreas
- Causes renal pain, reduced renal function, high blood pressure, renal stones, 50% of those affected get renal failure ≤ 60 years
- ~ 1 in 1,000 – 2,500 affected (PKD Charity)
- Genomic testing to help plan care pathways, for live donors and inform reproductive choices
- Treatments can reduce symptoms and risk of complications

Polycystic kidney disease (PKD)

East Genomics

- East GMSA working group to:
  - Identify key stakeholders – renal services, charities, patients and their families
  - Review current pathways and guidance
  - Define scope of project
  - February 2023 series of workshops focussing on pathways, competencies, context, equity of access

Feto maternal pathways

NHS East Genomics

- Fetal anomalies with a likely genetic cause – aneuploidy, whole exome, large panel, microarrays, copy number variations
- East of England audit planned: referral reasons and rates, test accuracy, outcomes, turn around times, failed or inconclusive results, equity of access
- 12 month project to understand how the test if offered, scope, gaps and challenges
- Denise Barnes presentation this afternoon

NICU/PICU

East Genomics

- 15% newborns admitted to NICU
- Initial clinical presentation may not predict outcomes
- Many investigations are invasive and sequential leading to delays in diagnosis and treatment
- Whole exome and genome sequencing available for acutely ill children with a likely monogenic disorder in NICU, PICU and other paediatric settings to give a more timely diagnosis to inform care and treatment pathways more accurately

NICU/PICU

NHS East Genomics

- Consent for genomic testing is complex and should include that the results may predict future health as well as diagnosis, scope and limitations, possibility of additional, unexpected or incidental findings, outcomes may be uncertain or unclear
- Workforce transformation pilot study:
  - NUH – genetic counsellors
  - CUH – registered nurses
  - UHL – genomic practitioner to be recruited

Lynch syndrome

NHS East Genomics

- Inherited cancer predisposition syndrome that increases risk of developing bowel, endometrial, ovarian, gastric, pancreatic, small bowel and renal cancers
- Work in progress to improve pathways for patients diagnosed with bowel and endometrial cancer to identify those at higher risk of Lynch syndrome
- Vicki Keisel and Leanne Barratt presentation this afternoon

Prostate cancer

East Genomics

- 1 in 8 men will get prostate cancer
- Men at higher risk:
  - Over 50
  - Black
  - First degree relative affected
- NHS England funded project United Against Prostate Action
- Genomic testing on the tumour to inform treatments and whether at increased of inherited predisposition
- Gemma Gunn presentation this afternoon

Monogenic diabetes

East Genomics

- Rare type of diabetes caused by single gene variation – diagnosis < 6 months, family history of maturity onset diabetes of the young or any type diagnosed at young age, diabetes doesn't fit type 1 or type 2
- ~2% of diabetes
- Genomic testing needed to identify the most appropriate treatment

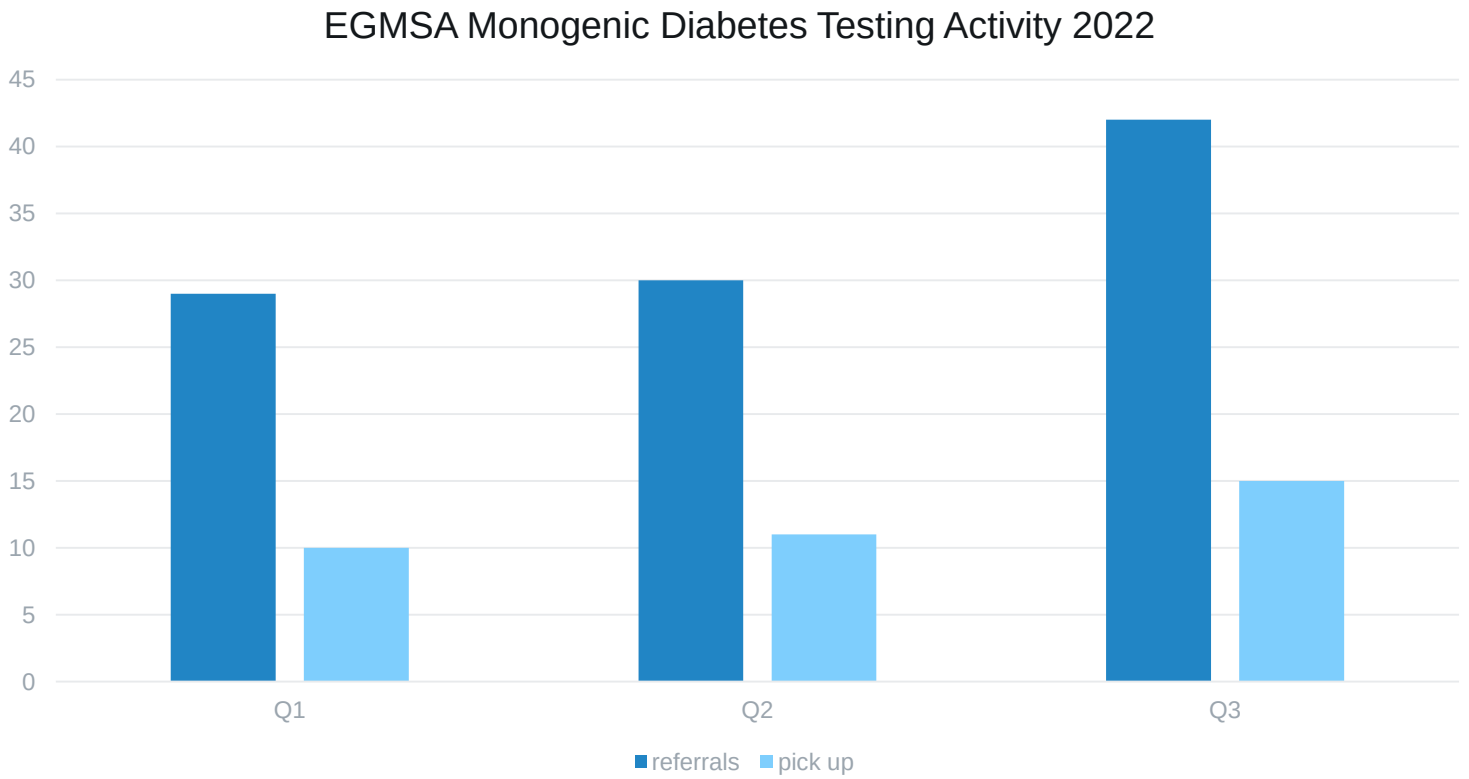
Monogenic diabetes

East Genomics

- HEE funded a national project to support genetic diabetes nurses
- GMSA supported project through clinical leadership and engagement with senior medical and nursing colleagues across the region
- Trained at least 1 adult and paediatric nurse and doctor in each relevant Trust – increased referral and diagnoses, reduced variation across NHS Trusts and improved outcomes
- GMSA supports Regional Monogenic Diabetics Network every 2 months

East GMSA Monogenic Diabetes Testing Activity

East Genomics



Familial hypercholesterolemia

NHS East Genomics

- Inherited condition which can cause very high cholesterol levels
- Untreated, increased risk of heart disease at young age
- ~1 in 250 affected
- Pilot project – Norwich area, CNS developing a hub model to coordinate service rather than current service at individual GP practice level

Nursing and midwifery genomic link forum

NHS East Genomics

- Meets every 2 months for an hour over Teams to share information and updates
- Links cascades information across their Trust to relevant nurses and midwives
- Identifying nurse and midwife specialists to participate in national work streams to identify local pathways, determine where genomics should be embedded to increase equity of access and reduce unwarranted variation and inform nurses and midwives' roles in this

East GMSA
Nursing and midwifery team

NHS East Genomics

Melissa Cambell-Kelly
Associate Nurse

melissa.cambellkelly@nnuh.nhs.uk

Annette Breen
Nurse Lead

annette.breen@nuh.nhs.uk

Vicky Carr
Nurse Lead

victoria.carr6@nhs.net

Katy Blakely
Regional Programme Manager - NNUH

katy.blakely@nnuh.nhs.uk

Break and networking

30 mins

#EastGenomicsNurseMidwife

The power of recommendations: examining presence and absence of shared first language in the antenatal clinic.

Professor Alison Pilnick (University of Nottingham)

East GMSA Nursing and Midwifery Conference,
November 22nd, 2022

The University of Nottingham

Introduction

- Collaborative project, ongoing since 2009 and spanning several grants, to compare aspects of antenatal screening practice in the UK and HK.
- Body of existing sociological work highlighting the impact that professional/client interaction can have on testing decisions (e.g. Williams et al 2005; Reid et al 2009; Pilnick 2008).
- Much of this work is interview based and/or conducted in largely mono-lingual settings.
- Our work uses audio/video recordings, and a conversation analytic approach.

Background

- Healthcare encounters involving participants from diverse linguistic backgrounds are becoming increasingly common as a result of globalisation and migration.
- But as Lewis (2002: 194) notes, the lack of empirical data from multicultural consultations 'makes it difficult to document the extent of the challenges…and their effects on experiences'.
- Moss and Roberts (2005): language use and understanding is dynamic and context dependent: there is no obvious marker denoting a language barrier

CA research on decision making

- Studies in other health-related contexts have shed light on how the way in which professionals elicit decisions can influence client choice (e.g. Collins et al 2005; Toerien et al 2013).
- Also a small but growing DA literature examining 'intercultural' healthcare encounters, e.g. Roberts (2007; 2009)- these analyses do not assume language as a significant variable *a priori*.
- Findings challenge some of the received wisdom about good communication skills.

Sample and methods

- Corpus of 120 consultations video-recorded in Hong Kong. These include native and non-native English and Cantonese speakers originating from various parts of Asia, North and South America, Europe and Australasia.
- Consultations conducted between L1/L1 speakers, L1/L2 speakers, and L2/L2 speakers
- Conversation analysis as primary analytic method
- Key question: **How does the presence/absence of a shared first language impact on these consultations, in terms of delivery of results and decisions making?**

A note about L1/L2 categorisations

- L1/L2 distinctions are commonly made in the sociolinguistic literature, and in literature on language acquisition.
- This can gloss the complexity of language acquisition and contextual competence.
- Our HK professionals identified as L2 English speakers, but their fluency made their consultations with English speaking clients hard to distinguish from L1/L1 consultations.

Findings

- Women in this corpus of consultations are receiving unwelcome news (a raised screening result).
- Professionals are very alert to language issues, and the need to find a common and comfortable basis for both parties.
- This is sometimes oriented to explicitly at the outset of consultations, e.g. Extract 1.

Extract 1:

- (12 WXZ)
- 1. D: 普通話的? ((in Mandarin))
- *Mandarin?*
- 2. P: 呀:對! 普通話.
- *Ah: yes! Mandarin.*
- 3. D: 普通話講不通呀!
- *I am not fluent in Mandarin (ah)!*
- 4. P: 哦? (.) 英文-英文也可以
- *Oh? (.) English- English works too.*
- 5. D: 講英文.
- *So let's speak English.*
- 6. P: Okay.

‘Backstage discussion’

- **Extract 3: (16 GKR)** (L2/L2 English, with L1 French between couple)

- 981. P: Tu fais quoi?
- *What do you want to do?*
- 982. H: C’est comme tu (0.5) préfères
- *It’s what you (0.5) prefer*
- 983. (0.8)
- 984. P: Yeah:h ((turns to N)) could you do both?

Does L1/L2 affect delivery of results?

- We found a common ‘script’ for delivery of results:
- Naming and explaining the components of the test that have been undertaken.
- Giving the numerical result.
- Contextualising this (e.g. ‘a bit high’, ‘relatively high’)
- Presenting the need for a decision.

- Differences were in the subdivision of the information into smaller components (cf ‘chunking and checking’ Silverman at al 1998) and in shorter turn constructions.

Does L1/L2 impact on formulations used to present decision making?

- We found no systematic differences in the way decisions about further diagnostic testing were framed.
- Across all categories we found:
- *Offers* of further testing
- *Recommendations* for further testing
- *Option listing* (Toerien et al 2013)
- (sometimes these were used in combination).

- Presence or absence of shared language did not restrict the range of formulations.

How pregnant women respond

- Most significant difference in our data set in absence of shared L1 is how *recommendations* regarding testing are received by pregnant women, and the linguistic resources they bring to bear for responding and/or contesting.

Extract 3 (L1/L1)

- *.h the chance of*
- 33. bb有唐氏綜合症就講緊呢即係.h大約三
  the baby having Down syndrome .h is, that is .hh about three
- 34. 百個，三百五十個先會有一個嫁(.)
  hundred a- (.) just one per three hundred and fifty people
- 35. *(1.2)*
- 36. 明唔明白呀? (0.4) ((while looking at the woman)) 咁所以嚟講呢
  do you understand? (0.4) ((looking at the woman)) so to speak, it still is
- 37. 都係比較高d嫁喇(.)咁所以我哋呢都會建議
  relatively higher (.) so, we recommend
- 38. 你係抽胎水。(0.7).h即係其實
  you to have amniocentesis. (0.7) .h that means actually,
- 39. P: em:: 都考慮咗我 (0.3)應該
  em:: um:: you have already considered my (0.3) .h it is probably
- 40. 唔係因為我個風濕性關節炎,
  not because of my rheumatoid arthritis?=
- 41.D: [我諗唔關事[°喇°
  =I think this is not related [then]
- 42. P: [吓]
  [I see]

Interrogating the evidence

- In this extract, the woman does not initially engage with the recommendation, but seeks further information about the reliability of the test.
- Although she frames this tentatively, with a negative polarity (Raymond 2003) and an orientation to medical expertise (ten Have 1991; Heath 1992), she still interrogates the evidence produced by the screening test before considering a decision.

How women respond (2):

- Examining receipt of recommendations in L1/L2 consultations shows a different picture.
- Women's responses are minimal.

Extract 4:

- 160. N: [係喇。].h咁呢個報告哩就變左建議你去做<嗰個>進一步
  [Right.] .h the report turns out to recommend you to have <that> further
- 161. 既檢驗啦。
  investigation,
- 162. P: 唔
  Hmm.
- 163. N: 咁因為你依家都十八週啦,
  because you are now at eighteen weeks,
- 164. P: 唔
  Hmm.
- 165. N: .h 咁如果要做進一步既檢驗，入侵性既檢驗呢，就
  .h if you want to do further investigation, invasive investigation, then
- 166. 要做羊水既。
  ((you)) would have to take amniocentesis.
- 167. P: 唔唔↓唔↑唔
  Umum ↓um↑um.
- 168. N: 咁囉。.h咁呢個係建議架啫。始終做唔做都係
  That's it. .h it is only a recommendation. To do it or not in the end is
- 169. 由你同先生決定既。
  decided by you and your husband.
- 170. P: 唔唔
  Mmm.

Extract 3 vs Extract 4

- These two extracts show a pattern common across the dataset.
- Women using L1 are better able to engage with recommendations, and less likely to treat them as definitive.
- Women using L2 are more likely to produce minimal responses and go along with what is recommended.
- Resisting a recommendation requires sophisticated linguistic resources (there is an interactional preference for acceptance (Pillet-Shore 2017)).

Alternative approaches?

- Recommendations are common in our data but are not the only formulation used.
- Examining *offers* shows a different pattern.
- Extract 5 (L2/L2)

Extract 5 (L2/L2)

- 30. D: .h so um it's up to you (.) We- we- we still call it
- 31. screen positive. (.) Because there is a small possibility
- 32. that the baby may have Downs.
- 33. (0.2)
- 34.P: Um hmm.
- 35.D: .hh Em but it's up to you whether you want the amniocentesis
- 36. (.) em: to check if [the baby]=
- 37.P: [is it the-]
- 38.D: = has Downs.
- 39.P: the one that put inside?
- 40. (0.2)
- 41.D: Yes, yes. ((while nodding))
- 42.P: Hhh ((nasal laugh)) I've discussed it with my husband. (.)
- 43. He said he doesn't want. (.) Heh heh=
- 44.D: =Right. ((while nodding))

How women respond (3)

- Offers require an active expression of choice
- They provide space for a pregnant woman to articulate a viewpoint.
- Option listing which does not conclude with a recommendation fulfils the same function.
- Where we see only minimal responses, it doesn't necessarily follow women have had their choices determined, but we can't see whether they're exercising choice or not.

Conclusions

- Professionals are sensitive to the contingencies of dealing with L2 clients.
- Whether, or how strongly, a particular course of action is promoted does not seem to depend on presence or absence of shared L1.
- But L2 women are more likely to make minimal responses, and hence to go along with recommendations
- L1 women may also go along with recommendations in the end, but do so with more active involvement.

Implications

- For practitioners who want to maximise engagement in decision making, recommendations may not be the best way to do this.
- Resisting a recommendation can be interactionally complex (Cederberg (2013) and self-representation in L2)).
- Offers of testing, or option listing which does not conclude with a recommendation, may be preferable.
- (As our data showed, L1/L2 is a simplistic distinction; what matters practically is whether language ability maps to context).

Genomics Education Programme

NHS
Health Education England

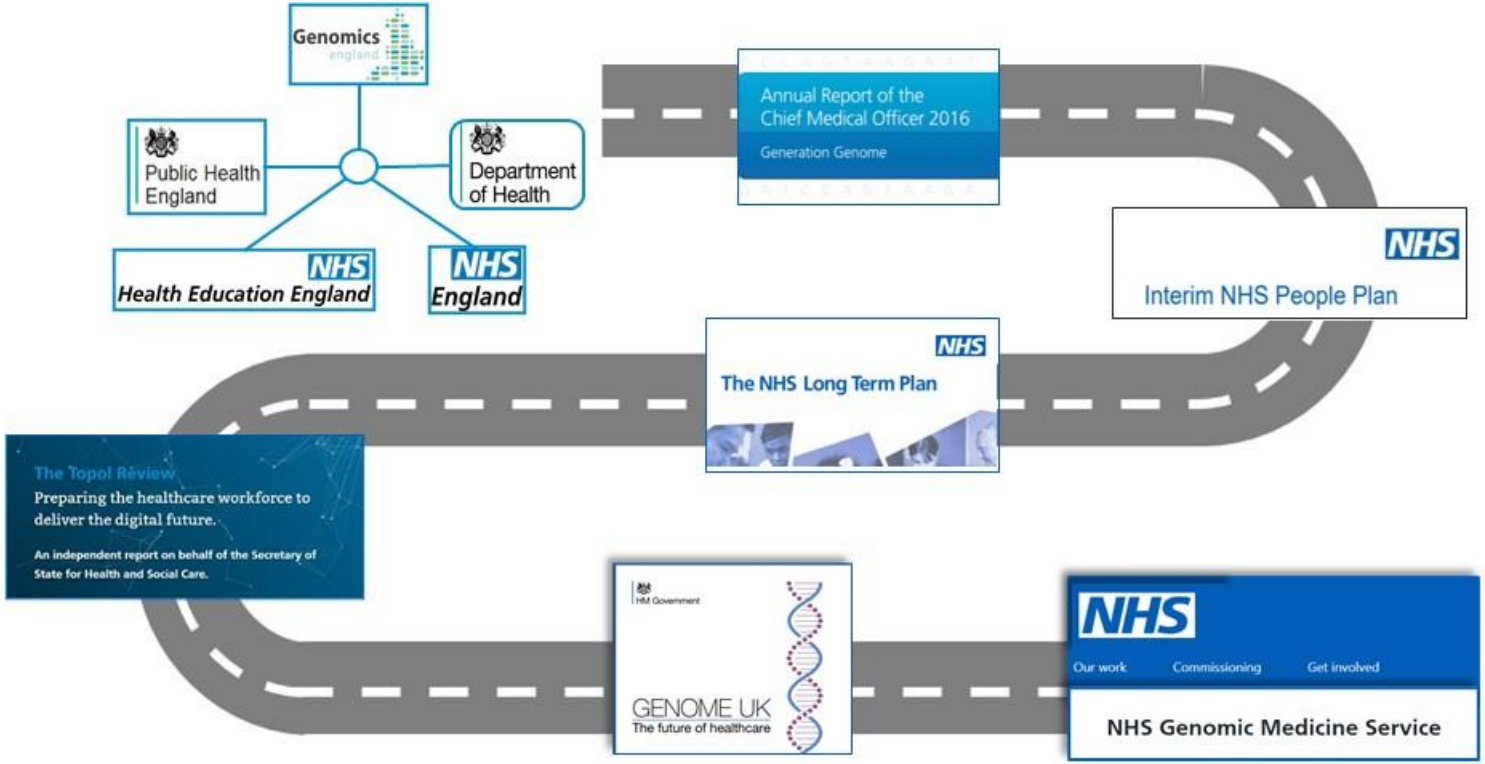
The Genomics Education Programme



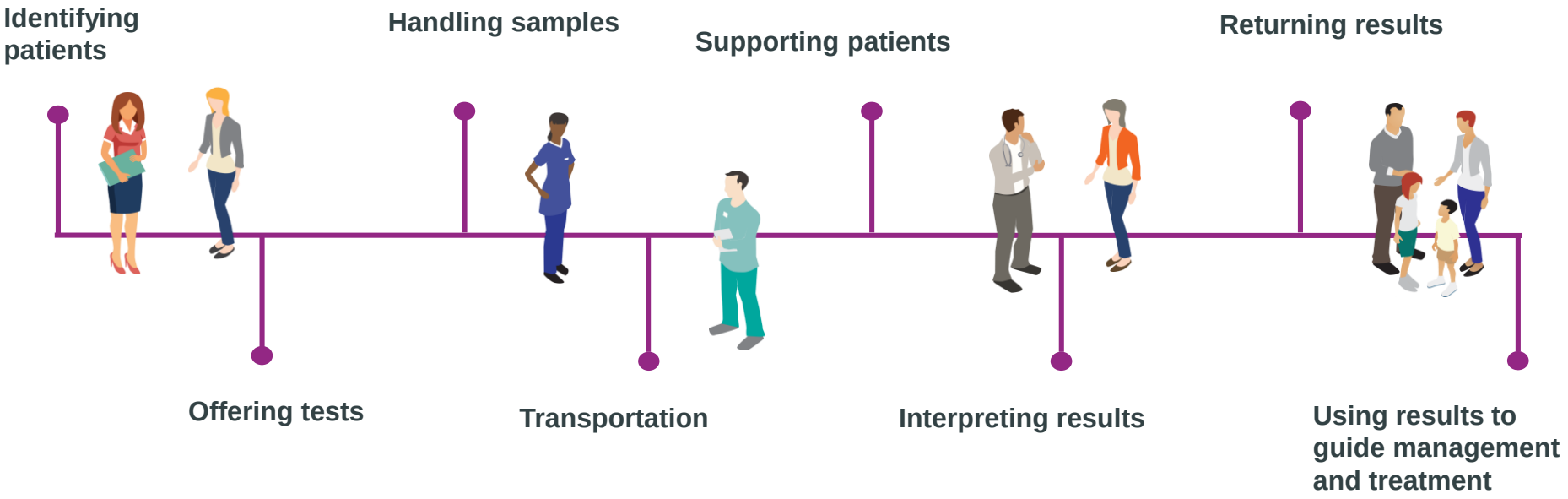

Ed Miller, Education Specialist, Genomics Education Programme, Health Education England

Edward.miller@hee.nhs.uk

Genomics in the NHS



Genomics will, to some extent, impact on many NHS healthcare professionals, however, **not everyone needs the same knowledge and skills** in genomics.



Education and training

“This will drive further workforce development and new education and training approaches to help embed genomics and the more detailed understanding of the influence of the genome on health, disease and personalised treatment”

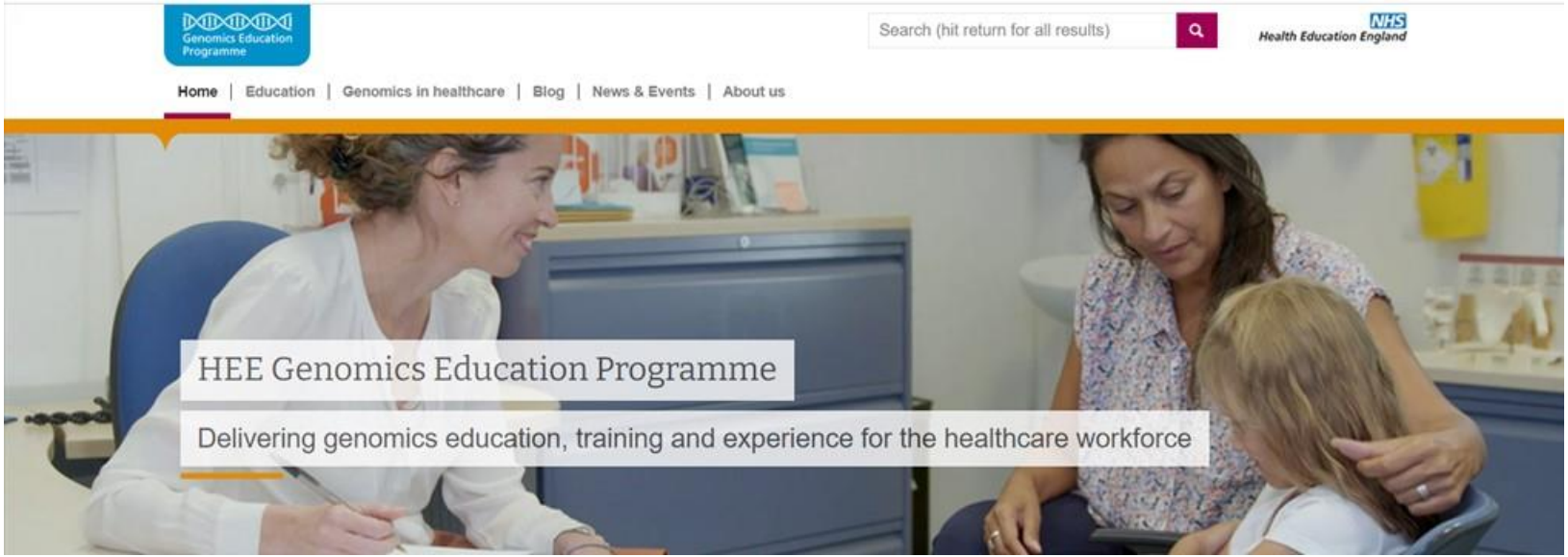
“There will be an ongoing requirement for genomics education to be built into the academic and training pathways of the whole healthcare workforce.”

“It must evolve from its solid foundations to a service that is embedded across the NHS care continuum, including primary and community care, with education and training at all levels to arm the workforce with up-to-date knowledge of genomics in their field.”

Education and training

Genomics has been incorporated into key training standards and curricula.

Health Education England's Genomics Education Programme (GEP)



HEE GEP key facts

Genomics Education Programme (GEP) **established in 2014**, with DHSC funding until 2017/18. HEE ‘business as usual’ from 2018.

GEP core objective: to ensure NHS workforce has the **knowledge**, **skills** and **experience** to deliver genomic medicine for patient benefit.

GEP audience: all **1.2 million** NHS professionals in England across the professional spectrum, from nursing associates to senior consultants.

Initial focus on supporting **100,000 Genomes Project**; now closely working with NHSE/I on roll-out of landmark **Genomic Medicine Service**.

Identifying workforce needs

Define training requirements mapped to roles, specialities and professions.

Educate and develop the NHS workforce

Resources tailored to needs and interests. Proactive and reactive learning.

Increase awareness of genomics across healthcare

Work with our networks to demonstrate the importance genomics will bring to patient care.

Build and join networks across the country

Ensure a nationally coherent approach to workforce development and training.
‘Do once and share’

Identifying workforce needs

- Dependent on **situation**

Directive (top down)

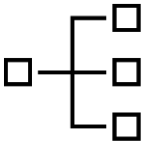
- to meet urgent need and gaps, e.g. resources for the GMS.
- respond to professional and regulatory bodies.

Consultative (bottom up)

- to allow workforce to define their learning needs and styles.

Identifying workforce needs: Clinical Pathway Initiative (CPI)

- introduce a **unified approach** to the integration of genomic medicine across different specialties;
- **identify** the workforce development and education needs;
- **harness** and **share** expertise from around the country;
- **avoid duplication*** of effort around resource development; and
- provide a '**bite-sized' clinically relevant** approach to genomic medicine
- Competency rather than profession led



FH Clinical Pathaway

Steps	Case-finding search in Primary Care IT system	Identifying relevant information to facilitate clinical diagnosis and eligibility for testing (EHR review)	Identifying relevant information to facilitate clinical diagnosis and eligibility for testing: Direct Patient Contact	Determine eligibility for testing
	1. Describes available informatics tools to support case-finding (e.g. CDRC, UCL-P or FAMCAT)	1. Demonstrates up-to-date knowledge of FH, including inheritance and clinical presentation)	1. Interprets diagnostic criteria to identify further information needed (e.g. family history, tests to confirm or exclude secondary causes)	1. Applies knowledge of diagnostic criteria and utilises relevant resources and patient information to enable decision-making
	2. Demonstrates ability to access, upload, run and generate output from case-finding tool within Primary Care IT system	2. Demonstrates up-to-date knoweldge of criteria for diagnosis (Clinical diagnostic criteria: Simon-Broome and Dutch Lipid Clinic Network, Genomic Test Directory: FH testing eligibility criteria)	2. Elicits relevant family history information (up to SDR and ages affected with symptoms or signs)	2. Assesses where FH genomic testing is appropriate in the patient's clinical pathway with reference to relevant pathways and guidelines (local/regional/national)
Identify resources	Specifc to case-finding tool: CDRC, FAMCAT, UCL	1. GeNotes (in progress)	1. GeNotes (in progress)	1. GeNotes (in progress)
	Resource (2)	2. RCGP Modules	2. RCGP Modules	2. RCGP Modules
	Resource (3)	3. Heart UK: Identifying FH in Pirmary Care	3. Heart UK: Identifying FH in Pirmary Care	3. Heart UK: Identifying FH in Pirmary Care
	Resource (4)	4. University of Northumbria course	4. University of Northumbria course	4. University of Northumbria course
Identify workforce	**Non-clinical, clinical or informatics**	**Primary healthcare professional: Pharmacist, GP, Practice Nurse, ANP, PA**	**Primary healthcare professional: Pharmacist, GP, Practice Nurse, ANP, PA (with expert advice if needed)**	**Primary healthcare professional: Pharmacist, GP, Practice Nurse, ANP, PA. Secondary Care: FH specialist nurse, Lipidology, Cardiology**

Identifying workforce needs: Clinical Pathway Initiative (CPI)

What it isn't

- A patient pathway
- Dictating how genomic medicine should be delivered
- Unadaptable, set in stone method

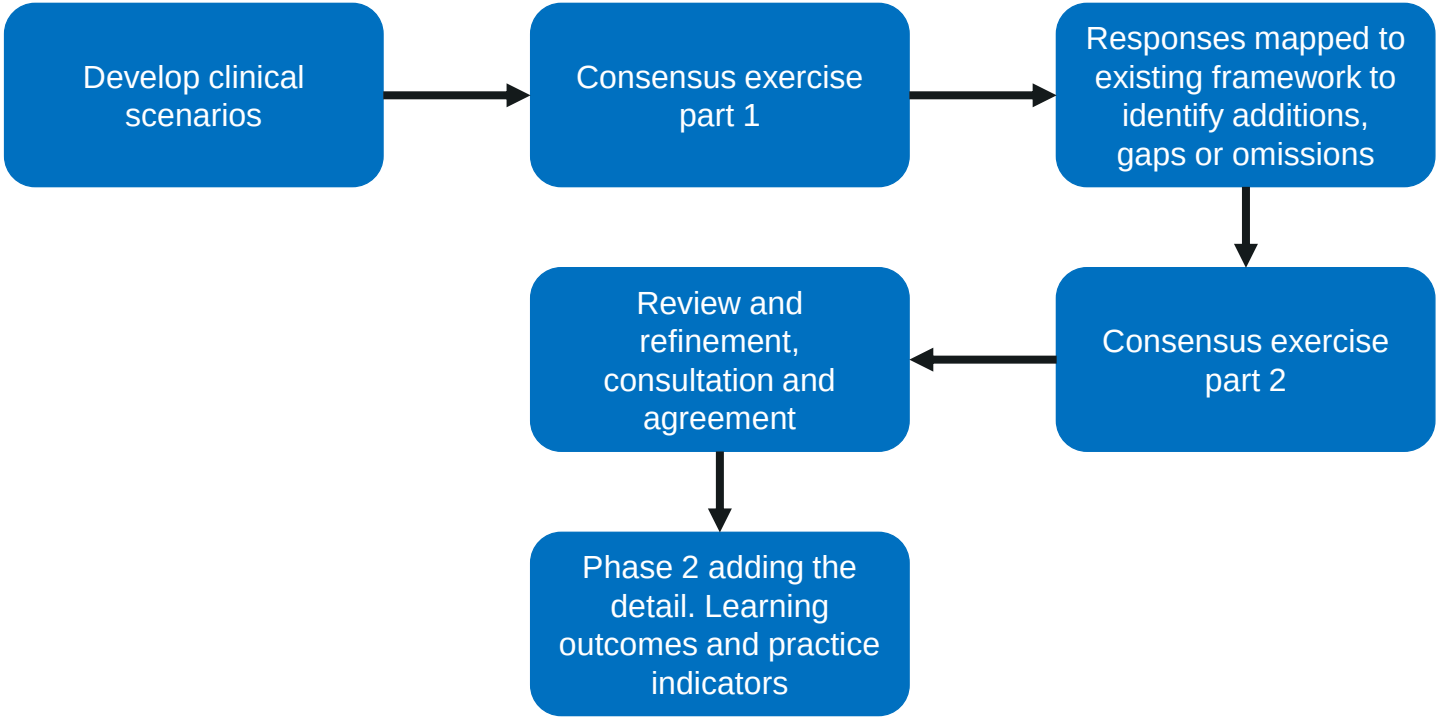
What it is?

- A step-by-step method to identify workforce development and education needs aligned to patient pathways across the NHS Genomic Medicine Service
- High level overview of clinical pathways
- An adaptive, flexible method

Identifying workforce needs: Competency Frameworks

- Profession agnostic
- Profession specific
  - **Nursing competency framework**
    - Building on previous work by Kirk et al 2014
    - For current and future workforce reflecting a modern genomics medicine service
    - Developed via a consensus method overseen by a steering group with representation from nursing education, practice and policy

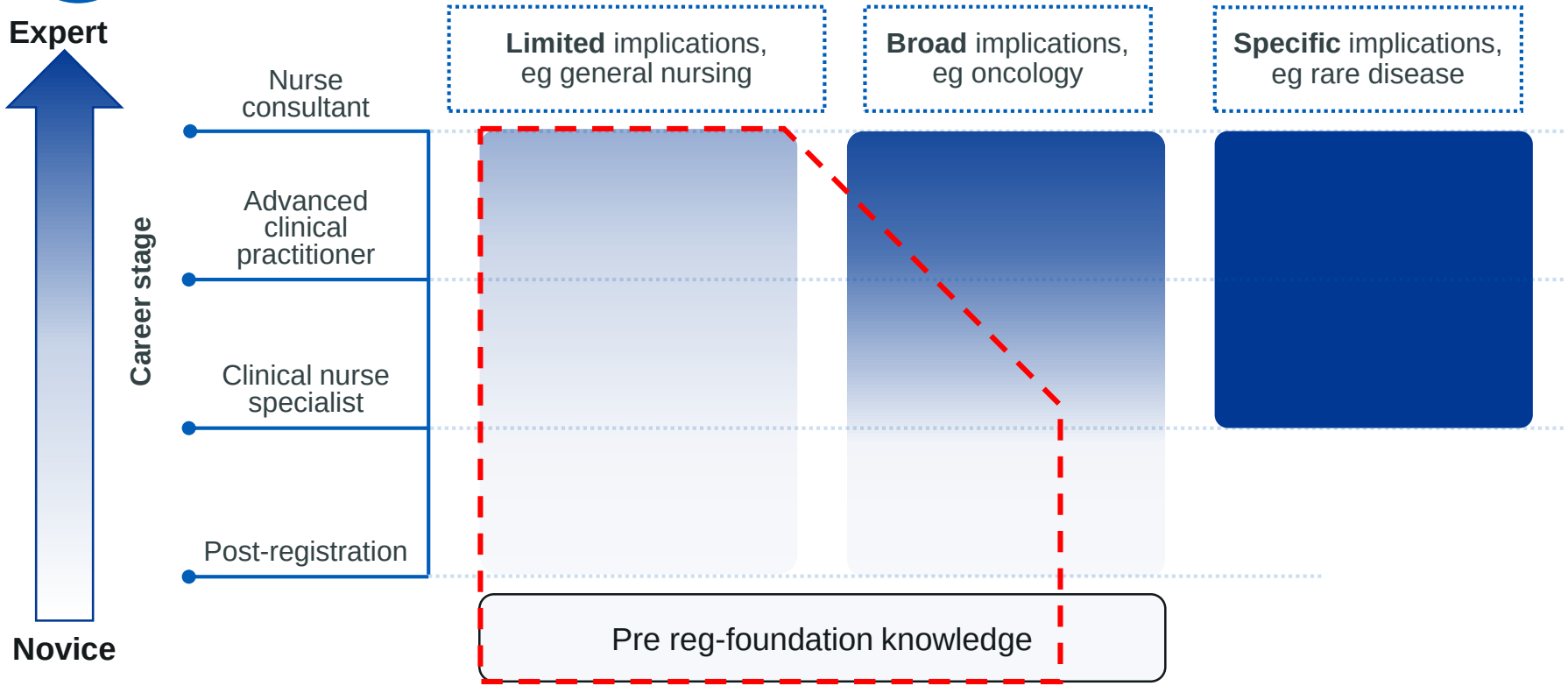
Identifying workforce needs: Competency frameworks



Identifying workforce needs: Competency frameworks

Scenario	Comp 1 Identification	2 Tailor information	3 Informed decision making & voluntary action	4 Role of genetics and genomics, and core knowledge	5 Testing and ethical legal and social implications	6 Own competence	7 Access and communicate information for self and others	8 Ongoing care
Child with secondary cancer	✓	✓	✓	✓	✓		✓	✓
Direct to consumer testing	✓	✓	✓	✓	✓	✓	✓	✓
Neonatal Diabetes	✓	✓	✓	✓	✓	✓	✓	✓
Ovarian	✓	✓	✓	✓	✓	✓	✓	✓
Point of Care testing	✓	✓	✓	✓	✓	✓	✓	✓
Trio Exome	✓	✓	✓	✓	✓	✓	✓	✓

Identifying workforce needs: Competency frameworks



Educate and develop the NHS workforce

Novice

Bitesize

Genomics 101

GMS courses

FutureLearn course

CPPD modules

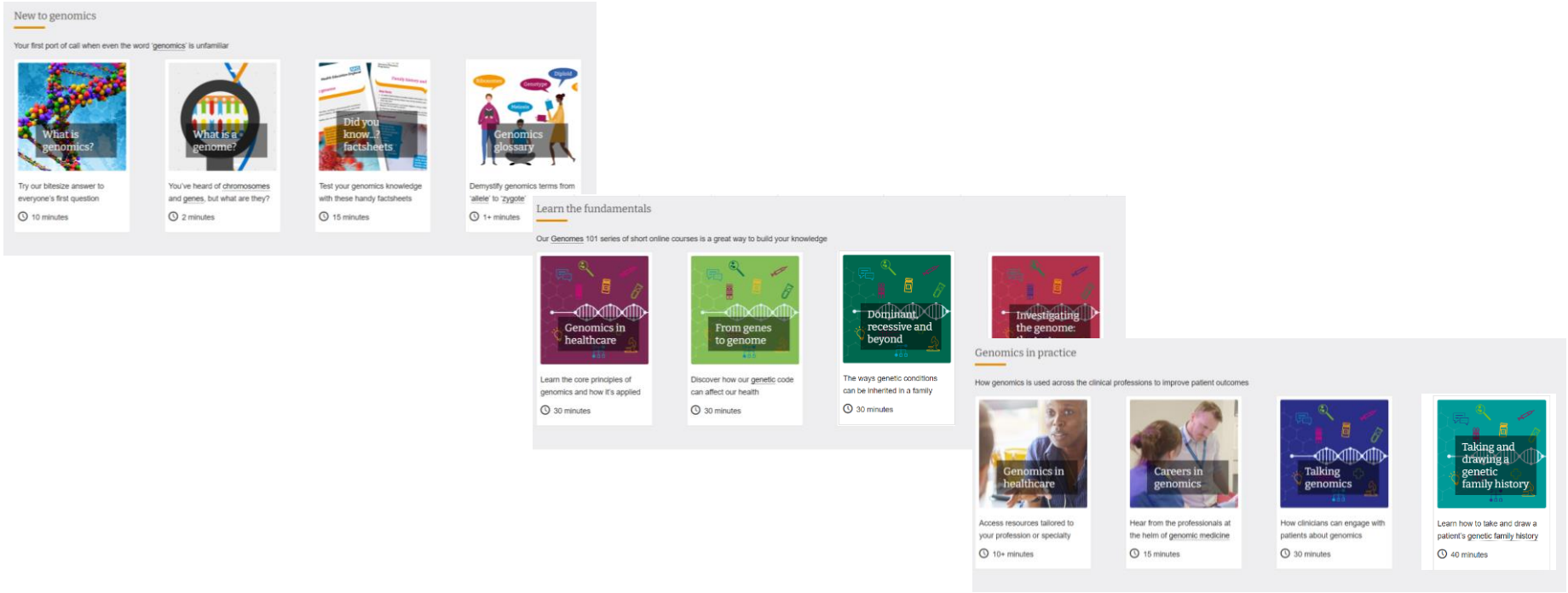
Master's in Genomic Medicine

Expert

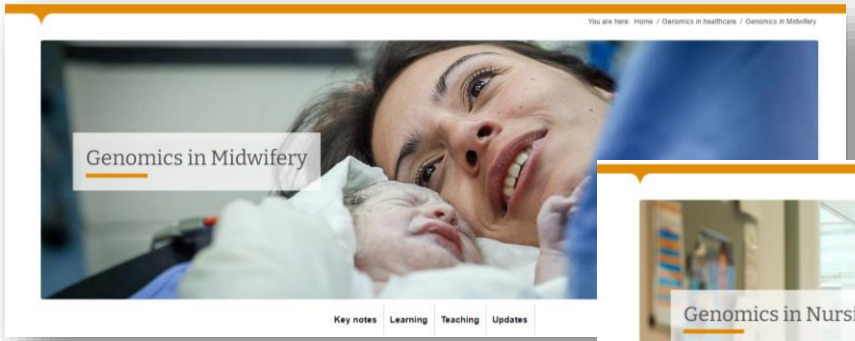
Awareness raising: Videos, blogs, social media

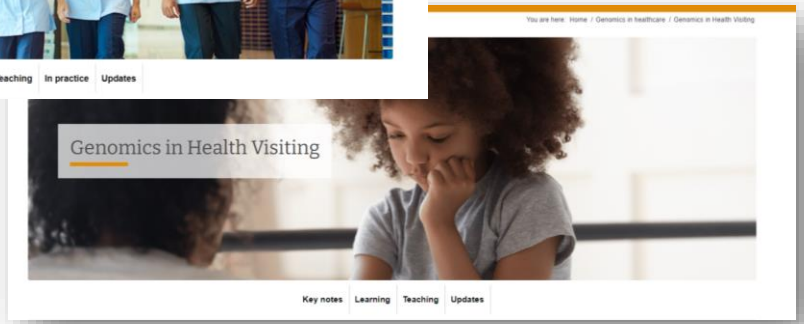
Based on Benner, P.(1982)

Educate and develop the NHS workforce: Learning journeys

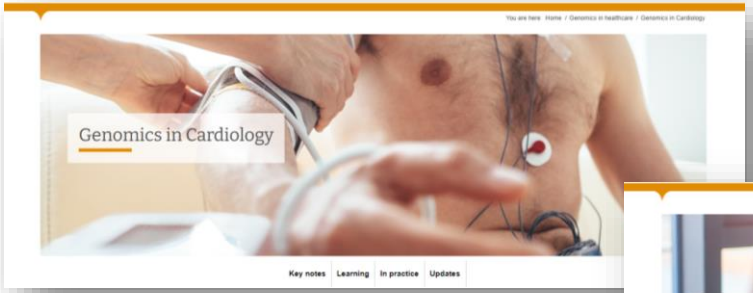


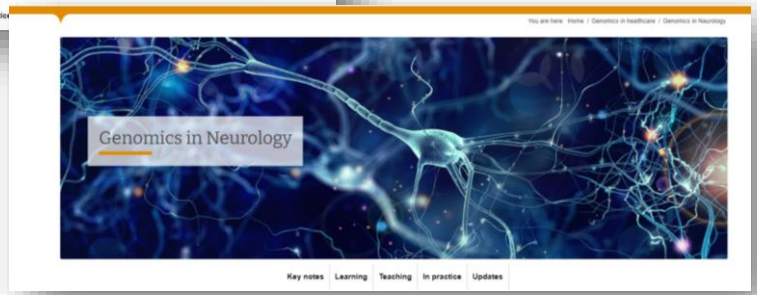
Educate and develop the NHS workforce: Webpages





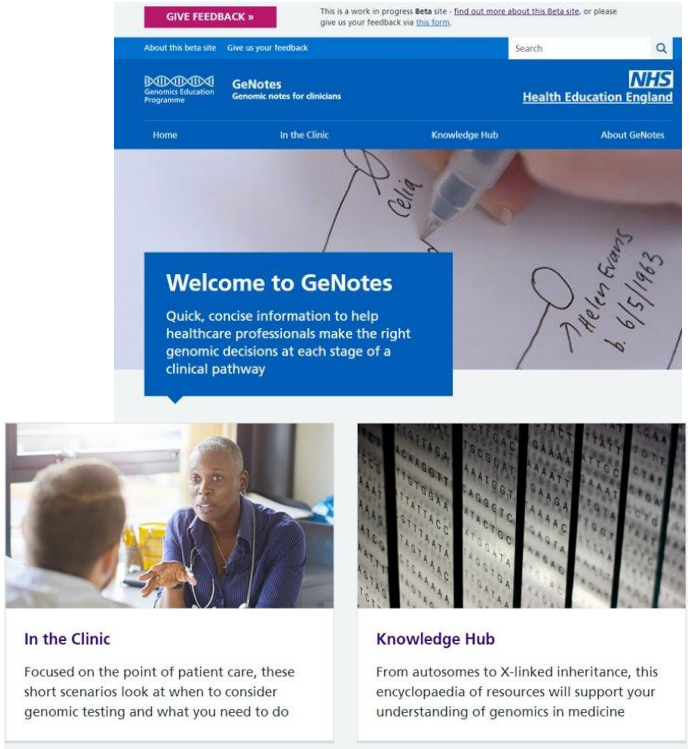
Educate and develop the NHS workforce: Webpages





Educate and develop the NHS workforce: GeNotes

- Two-tier **“just in time”** online resource.
- Aligns to national priorities
- Developed **by** mainstream clinicians **for** mainstream clinicians
- Aims to **support the clinician at the point of need** and provide opportunities for extended learning.
- **Iterative**: easily added to/updated
- **Collaboration** between
  - HEE Genomics Education Programme
  - Academy of Medical Royal Colleges; and
  - Individual Royal Colleges
- **Free** to access for all NHS staff



Educate and develop the NHS workforce: Educator's Toolkit

*2.2 Demonstrate knowledge of epidemiology, demography**, genomics** and the wider determinants of health, illness and wellbeing at all stages of life and apply this to an understanding of patterns of health and illness and health outcomes.*

*3.2 Demonstrate and apply knowledge of body systems and homeostasis, human anatomy and physiology, biology, **genomics**, pharmacology, social and behavioural sciences, to inform accurate nursing assessments and develop appropriate person-centred care plans.'*

*2.3 demonstrate the principles of epidemiology, demography**, genomics** and how these may influence health and wellbeing outcomes*

*3.2 Demonstrate and apply knowledge of body systems and homeostasis, human anatomy and physiology, biology, **genomics**, pharmacology, social and behavioural sciences, when delivering care*

Educate and develop the NHS workforce: Educator's Toolkit

- Supporting educators with the incorporation of genomics into curricula.

- The toolkit aims to demonstrate that genomics:
  - is not a standalone subject;
  - can be knitted throughout curricula;
  - is applicable to different levels of teaching, nursing fields and areas of practice.

Educate and develop the NHS workforce: Educator's Toolkit

- Case-study based, categorised by:
  - Nursing field e.g. Adult, CYP.
  - Nursing activities e.g. Identification, communication, management.
  - Area of practice e.g. rare disease, cancer.
  - NMC platform and outcomes.
- Each case study is supported by additional teaching guidance and suggestions, in addition to further learning opportunities.

Educate and develop the NHS workforce: Educator's Toolkit

- Interpretation of the standards

Platform 1: Being an accountable person	
Outcomes	Genomics in Practice
1.9 understand the need to base all decisions regarding care and interventions on people's needs and preferences, recognising and addressing any personal and external factors that may unduly influence their decisions	If a genomic condition is suspected, consider impact on family (sometimes community), culture, religion, prior experience and personal values.

Platform 3. Assessing needs and planning care	
Outcomes	Genomics in Practice
3.5 demonstrate the ability to accurately process all information gathered during the assessment process to identify needs for individualised nursing care and develop person-centred evidence-based plans for nursing interventions with agreed goals	Patient family histories (genetic conditions) and genomic information that may impact on treatment and care (pharmacogenomics) of individual or other family members.

Bill's story

+ Identification and diagnosis
+ Treatment and management
+ Raising awareness
+ Specialist services
+ The role of nurses
+ Supporting families

www.genomicseducation.hee.nhs.uk/nursing-educators-toolkit/

AT-A-GLANCE

Clinical focus: Cardiology, common conditions

Nursing activities: Identification, management and ongoing care, communication, family care, education

NMC platform and outcomes: 1 (1.8, 1.9, 1.13, 1.18); **2** (2.2, 2.4, 2.5, 2.9, 2.10); **3** (3.1, 3.2, 3.5, 3.11, 3.12, 3.15, 3.16); **4** (4.2, 4.3, 4.5, 4.15); **5** (5.4); **7** (7.1, 7.8)

Teaching moments

Paragraph 1 | What is meant by a dominantly inherited condition?

Paragraph 2 | What is a family history and why are they important?

Paragraph 3 | What genetic tests exist that can look at more than one gene?

Paragraph 10 | What is meant by cascade testing?

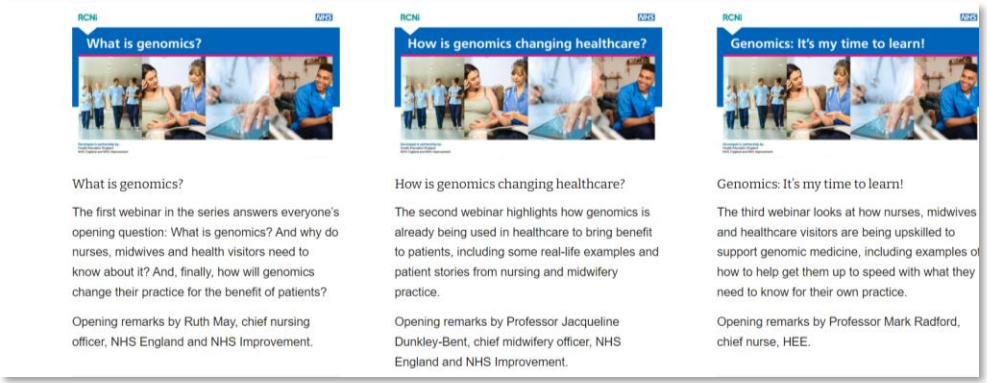
Discussion points

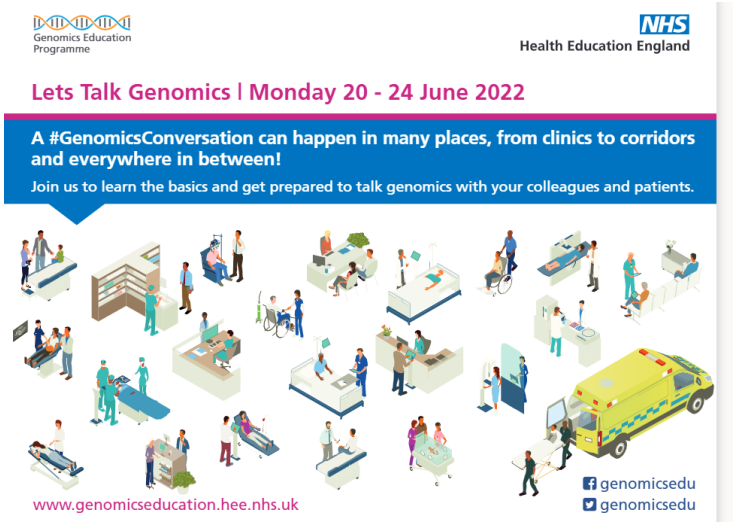
Paragraph 2 | Consider common clinical indicators or red flags when identifying genetic conditions, for example, family history, early onset of condition, symptoms in young.

Paragraph 5 | Even though the condition is genetic, it can be managed. How does this challenge the misconception that there's nothing that can be done about genetic conditions?

Paragraph 7 | What are the benefits of having an understanding awareness of this condition?

Increase awareness of genomics across healthcare





Build and join networks across the country

HEE via Genomics Education Programme

Stakeholder networks

- GLH/GMSA Workforce Network
- Primary Care Network
- HEI Network

NHSE Transformation Projects

- Pharmacy
- Nursing and midwifery
- Medical

External networks

- NHSE Genomics Unit
- Genomics England
- Workforce Redesign Partnership Board
- AoMRC
- Third sector (eg Macmillan)
- AHSN
- National School of Healthcare Science

Strategies

- NHS Long Term Plan
- Interim People Plan
- The Topol Review
- HEE Cancer Strategy
- UK Rare Diseases Strategy
- Life Sciences Strategy

Build and join networks across the country

- Joint workforce steering group
- Royal Colleges
- Third Party
  - Macmillan
  - iHV
- HEIs
  - MSc Framework
  - UWE: Genomic and Counselling Skills for Nurses and Healthcare Professionals

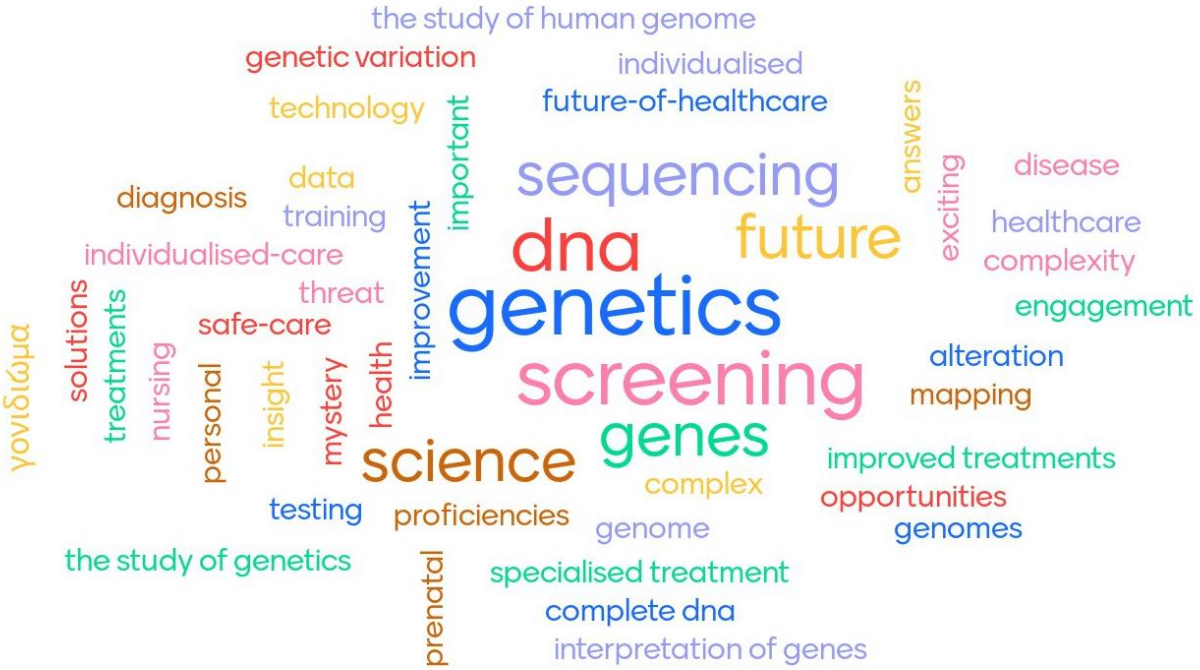
Ongoing challenges

- How much and what genomics to teach
- Demonstrating relevance to practice
- Lack of confident and experienced educators
- Time to learn

Final thoughts and take home messages

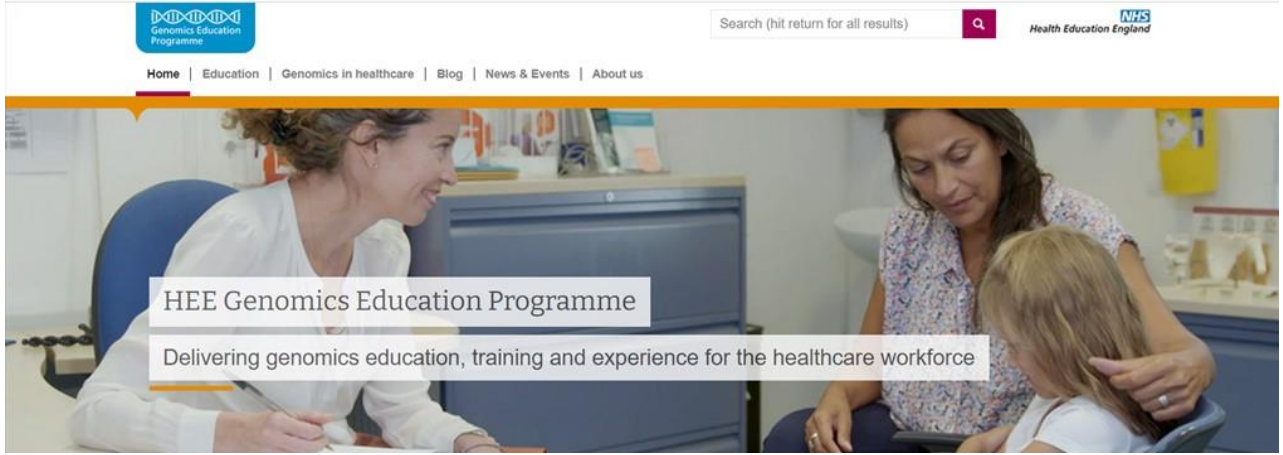
- Talk to 3 of your colleagues about genomics
- Take one of the Genomics 101 eLearning modules
- Do you have patient story or example you could share?
- Think genomics…it may not always be obvious

What does the word genomics mean to you?



What does the word genomics mean to you now?

Get in touch



www.genomicseducation.hee.nhs.uk

Edward.miller@hee.nhs.uk

@genomicsedu

Panel Q&A

Chaired by Emma Tonkin (20 mins)

Lunch

Reconvene at 1.30pm

#EastGenomicsNurseMidwife

Information stands:

NHS East Genomics

Genomics Education Programme

Genomics england

Parallel Sessions

Nursing – King's Hall (this room)

1.30pm: **Lynch syndrome; mainstreaming and patient story**
2pm: **United Against Prostate Cancer Project and patient story**
2.30pm: **Familial epilepsy and patient story**

Midwifery – Cromwell Room

1.30pm: **Genomics in Midwifery**
1.50pm: **Fetomaternal Pathways**
2.15pm: **Newborn blood spot screening: whole genome sequencing**
2.30pm: **Genomics and consanguinity; screening, testing and counselling**

Reconvene in King's Hall at 3.20pm

NHS East Genomics

Making a Difference:

Genomics in Nursing and Midwifery Conference

Afternoon Nursing Sessions

Venue Wifi:

Network: Conference

Password: Mercure24

Twitter:

#EastGenomicsNurseMidwife







NHS
East Genomics

Lynch syndrome

Vicki Kiesel, Lead Genetic Counsellor & East Midlands GMSA LS lead

Lynch Syndrome (LS)

NHS East Genomics

- An estimated 175,000 people in the UK have Lynch syndrome
- 1 in 350 are affected
- Leads to over 1,100 colorectal cancers a year in the UK.
- Responsible for 3% of CRC and Endometrial cancers
- Tumours usually show absence of 4 proteins: identified by immunohistochemistry (IHC)

What are the risks of Lynch Syndrome?

NHS East Genomics

Lifetime cancer risks:	
Colorectal	10-57%
Endometrial	13-49%
Gastric	13-19%
Ovarian	9-12%
Biliary tract	2%
Urinary tract	4%
Small bowel	1-4%
Brain/CNS	1-3%

Gene specific risks

East Genomics

Male *MLH1* approximate risks*

Cancer type	*MLH1* mutation carrier (up to 75)	Population lifetime risk
Colorectal	57%	7%
Endometrial	-	-
Ovarian	-	-
Upper gastrointestinal	22%	5%
Ureter/kidney	5%	3%
Urinary Bladder	7%	2%
Brain	<1%	<1%
Prostate	Similar to population/ may be increased	18%

Female *MLH1* approximate risks*

Cancer type	*MLH1* mutation carrier (up to 75)	Population lifetime risk
Colorectal	48%	6%
Endometrial	37%	3%
Ovarian	11%	2%
Upper gastrointestinal	11%	4%
Ureter/kidney	4%	2%
Urinary Bladder	5%	<1%
Brain	2%	<1%
Prostate	-	-

Highest risks
57% CRC: MLH1 males
22% upper GI: MLH1 males
49% EC: MSH2 females
17% OC: MSH2 females
24% prostate: MSH2 males

Male *MSH2* approximate risks*

Cancer type	*MSH2* mutation carrier (up to 75)	Population lifetime risk
Colorectal	51%	7%
Endometrial	-	-
Ovarian	-	-
Upper gastrointestinal	20%	5%
Ureter/kidney	18%	3%
Urinary Bladder	13%	2%
Brain	8%	<1%
Prostate	24%	18%

Female *MSH2* approximate risks*

Cancer type	*MSH2* mutation carrier (up to 75)	Population lifetime risk
Colorectal	47%	6%
Endometrial	49%	3%
Ovarian	17%	2%
Upper gastrointestinal	13%	4%
Ureter/kidney	19%	2%
Urinary Bladder	8%	<1%
Brain	3%	<1%
Prostate	-	-

Male *MSH6* approximate risks*

Cancer type	*MSH6* mutation carrier (up to 75)	Population lifetime risk
Colorectal	18%	7%
Endometrial	-	-
Ovarian	-	-
Upper gastrointestinal	8%	5%
Ureter/kidney	2%	3%
Urinary Bladder	8%	2%
Brain	2%	<1%
Prostate	Similar to population/ may be increased	18%

Female *MSH6* approximate risks*

Cancer type	*MSH6* mutation carrier (up to 75)	Population lifetime risk
Colorectal	20%	6%
Endometrial	41%	3%
Ovarian	11%	2%
Upper gastrointestinal	4%	4%
Ureter/kidney	6%	2%
Urinary Bladder	1%	<1%
Brain	1%	<1%
Prostate	-	-

- MSH6 & PMS2 have lower risks generally
- Endometrial is 41% MSH6

Male *PMS2* approximate risks*

Cancer type	*PMS2* mutation carrier (up to 80)	Population lifetime risk
Colorectal	13%	7%
Endometrial	-	-
Ovarian	-	-
Upper gastrointestinal	Similar to population	5%
Ureter/kidney	Similar to population	3%
Urinary Bladder	Similar to population	2%
Brain	Similar to population	<1%
Prostate	Similar to population	18%

Female *PMS2* approximate risks*

Cancer type	*PMS2* mutation carrier (up to 80)	Population lifetime risk
Colorectal	12%	6%
Endometrial	13%	3%
Ovarian	Similar to population	2%
Upper gastrointestinal	Similar to population	4%
Ureter/kidney	Similar to population	2%
Urinary Bladder	Similar to population	<1%
Brain	Similar to population	<1%
Prostate	-	-

Identifying those at risk

~95% undiagnosed

Even when a family has been diagnosed with Lynch syndrome they are not always managed appropriately.

Somatic Screening for Lynch syndrome

NHS East Genomics

Molecular testing strategies for Lynch syndrome in people with colorectal cancer

Diagnostics guidance

Published: 22 February 2017

www.nice.org.uk/guidance/dg27

Testing strategies for Lynch syndrome in people with endometrial cancer

Diagnostics guidance

Published: 28 October 2020

www.nice.org.uk/guidance/dg42

What is Somatic Screening for Lynch Syndrome?

NHS East Genomics

- Immunohistochemistry
- Stains the proteins for 4 genes
- Genes cause LS: MLH1, MSH2, MSH6, PMS2
- If the proteins are absent it is because:
  - Gene pathogenic mutation causing Lynch syndrome
  - Sporadic cancer
- 30% of EC and 15% of CRC shows abnormal IHC (MMR deficient)
- The next stage of testing (MLH1 and/or BRAF) differentiates between inherited and sporadic

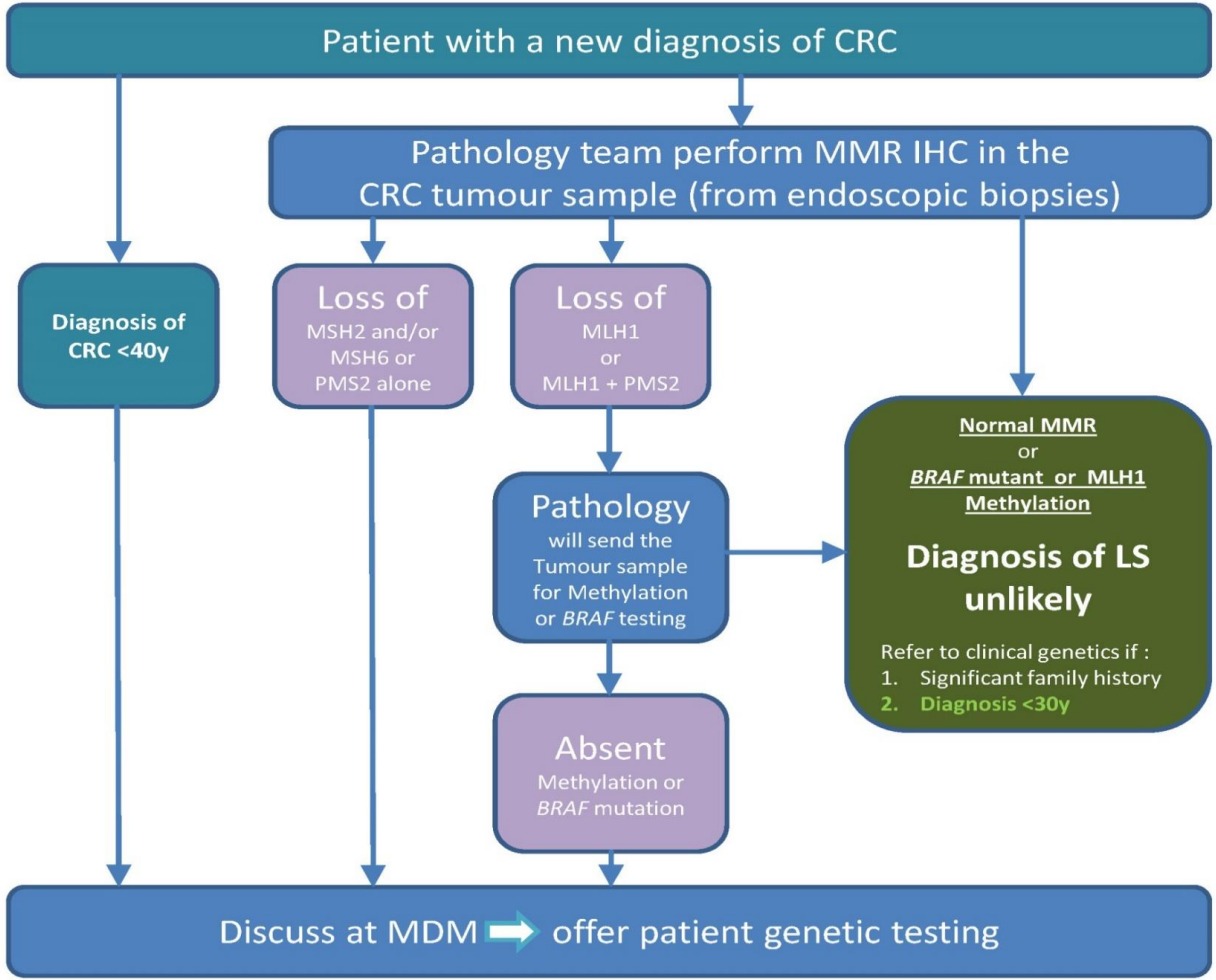
National and local audits suggest that only 1/3 of abnormal IHC/MSI results were actioned!

Quality Standards: QS20 Colorectal Cancer: Feb 2022: Statement 1 Testing for Lynch Syndrome

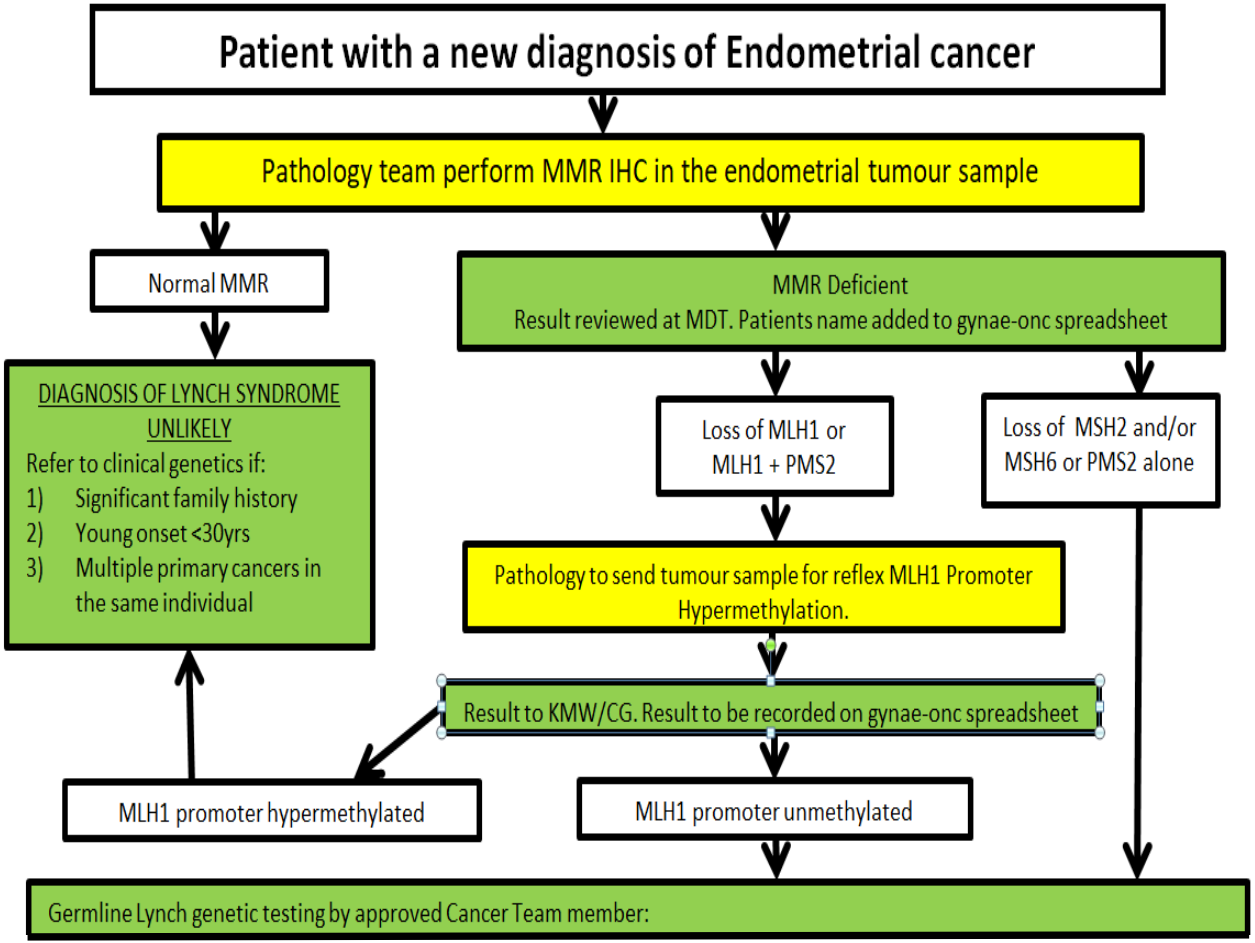
- Local LS lead should be identified
- HC needs to be performed on all CRC
  With reflex to BRAF / MLH1 Methylation where necessary
- Healthcare professionals must be aware of local protocols and can identify when to refer to clinical genetic services

NHS England

Testing for Lynch syndrome and offer of cascade testing for family members are included in the 2022/23 priorities and operational planning guidance from NHS England: "ensuring that every person diagnosed with colorectal and endometrial cancer is tested for Lynch syndrome (with cascade testing offered to family members)"



NHS
East Genomics

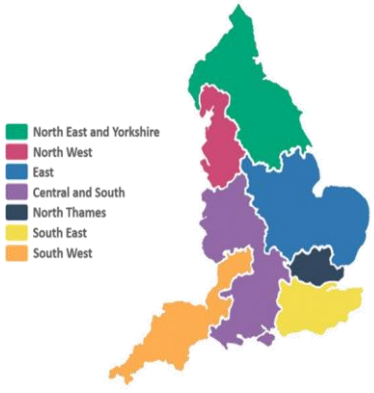


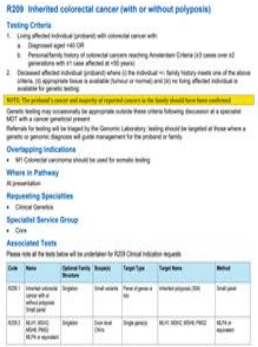
NHS
East Genomics

Pathway delivery is supported by

NHS East Genomics

- National NHSE Transformation Project led by North Thames and South East GMSAs
- National Oversight Group meet monthly
- Cancer Alliances
  - Sally Picken, EMCA
  - Genomic Expert Clinical Advisory Groups
- The new National Genomic Medicine Service
  - 7 supra-regional NHS Genomic Laboratory Hubs (GLHs)
  - 7 Genomic Medicine Service Alliances (GMSAs)
    - GMSA LS team
- The National Genomic Test Directory





R209 Inherited colorectal cancer (with or without polyposis)

Testing Criteria

1. Living affected individual (proband) with colorectal cancer with
   a. Diagnosed aged <40 OR
   b. Personal/family history of colorectal cancers reaching Amsterdam Criteria (≥3 cases over ≥2 generations with ≥1 case affected at <50 years)
2. Deceased affected individual (proband) where (i) the individual +/- family history meets one of the above criteria, (ii) appropriate tissue is available (tumour or normal) and (iii) no living affected individual is available for genetic testing

NOTE: The proband's cancer and majority of reported cancers in the family should have been confirmed

Genetic testing may occasionally be appropriate outside these criteria following discussion at a specialist MDT with a cancer genetics/ clinical present

Referrals for testing will be triaged by the Genomic Laboratory; testing should be targeted at those where a genetic or genomic diagnosis will guide management for the proband or family.

Overlapping Indications

- M1 Colorectal carcinoma should be used for somatic testing

Where in Pathway

At presentation

Requesting Specialties

- Clinical Genetics

Specialist Service Group

- Core

Associated Tests

Please note all the tests below will be undertaken for R209 Clinical Indication requests

Code	Name	Optional Family Structure	Group(s)	Target Type	Target Name	Method
R209.1	Inherited colorectal cancer with or without polyposis Small panel	Singleton	Small variants	Panel of genes or loci	Inherited polyposis, CRC	Small panel
R209.2	MLH1, MSH2, MSH6, PMS2 MLPA or equivalent	Singleton	Exon level CNVs	Single genes	MLH1, MSH2, MSH6, PMS2	MLPA or equivalent

When is Germline Genetics needed?

NHS East Genomics

- MMR deficient IHC showing:
  - Absence of any gene other than MLH1
- MMR deficient with absent MLH1 AND no BRAF (CRC only) or MLH1 methylation
- FHX questionnaire is NOT needed

What's the future?

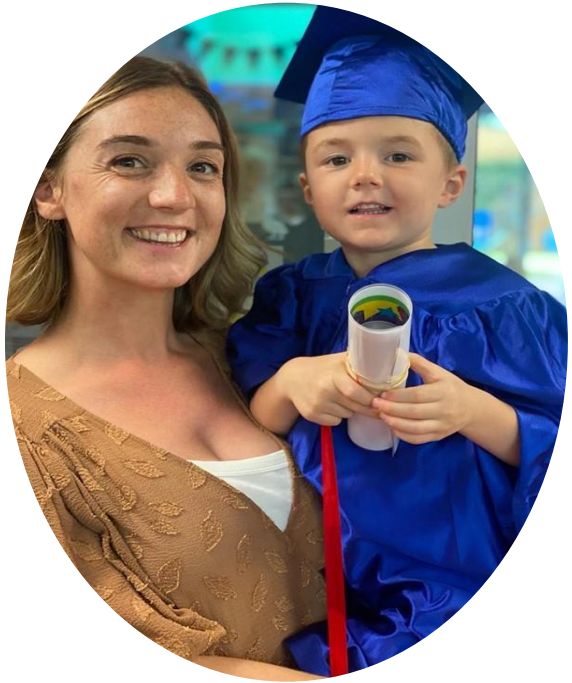
NHS East Genomics

- All patients with abnormal IHC will be discussed in MDTs
- Germline testing & consenting to occur in MDTs
- All LS patients to be referred to LS expert hubs
  - 1 Network across East Midlands: NUH and UHL
- All LS patients to be discussed by expert hubs to ensure appropriate management
- Colonoscopic surveillance to be arranged 2 yearly by national bowel screening program from April 23

NHS
East Genomics

Any questions?

East Genomics



Leanne Barratt

Patient

Leanne is a Lynch Syndrome patient, under the care of Julian Barwell in Leicester.

After discovering she had the MSH2 genetic malfunction back in 2010 she had a son, Jasper, in 2017 via PGD IVF to eliminate the risk of inheriting the gene.

She works for Next as a Brand Marketing Manager, working on photoshoots and creating advertising assets for the business.

NHS
East Genomics

Gemma Gunn

United Against Prostate Cancer Project and patient story

United Against Prostate Cancer

United Against Prostate Cancer

NHS East Genomics

- The United Against Prostate Cancer team are using a collaborative approach to tackling health inequalities in relation to prostate cancer across the East GMSA.

- This is through working with clinical and scientific colleagues, stakeholder partnerships and community champions.

- We are aiming to improve the care of men and support for their families by improving awareness of prostate cancer and genetic testing within high-risk groups, individuals with a relevant family history and/or of an African / African Caribbean heritage.

Why prostate cancer?

East Genomics

1 in 8	Will develop prostate cancer
1 in 4	Individuals of African heritage will develop prostate cancer
Most common male cancer	52,300 new prostate cancer cases in the UK every year
140	That's more than 140 every day
27%	Of male cancers in the UK are prostate cancer
12%	Is the projected increase in incidence rates of prostate cancer by 2035
100%	Almost all patients diagnosed with prostate cancer at stage 1 or 2 will survive their disease beyond 5 years
49%	Of patients diagnosed with prostate cancer at stage 4 will survive their disease beyond 5 years

For up to date statistics see Cancer Research UK website

https://www.cancerresearchuk.org/health-professional/cancer-statistics/statistics-by-cancer-type/prostate-cancer#heading-Zero

Prostate cancer can affect any individual who was identified as a male at birth.

For trans women who receive hormones, studies appear to show a reduction in the risk of developing prostate cancer, but the risk is not completely removed.

Genital reconstructive surgery does not remove the prostate, therefore prostate cancer is still possible after surgery.

https://prostatecanceruk.org/prostate-information/are-you-at-risk/trans-women-and-prostate-cancer

Identifying and understanding risk

NHS East Genomics

Individual who has/had prostate cancer	Increased Risk
Father	2.1-2.4
Brother	2.9-3.3
Second degree relative (grandfather, uncle, nephew or half sibling)	1.9

Known genetic mutation	Increased Risk
BRCA 2 <65years	7.33
BRCA 2 >65years	4.65
Lynch Syndrome	2.1-4.9

This risk increases further if an affected relative was diagnosed under 60 or there are more than one relative diagnosed with prostate cancer. A family history of breast cancer increases the risk of developing prostate cancer.

https://www.cancerresearchuk.org/health-professional/cancer-statistics/statistics-by-cancer-type/prostate-cancer/risk-factors#ref-2

Recognising health inequalities: Ethnicity

NHS East Genomics

1 in 4 Black men will develop prostate cancer

Twice as likely to develop prostate cancer than white men

The tumour is often a more aggressive type

Black men are twice as likely to die from prostate cancer then white men

UAPC approach to addressing the identified inequalities

East Genomics

Clinical Pathway

Community outreach

Access to testing

East Genomics

For a patient with prostate cancer to be eligible for germline testing, they must also have a Manchester Score >15.

As we have seen, an individuals prostate cancer risk increases by 2-3 times if a first or second degree relative has prostate cancer

A BRCA 2/Lynch syndrome mutation significantly increases prostate cancer risk

Yet, for a patient with prostate cancer with a family history only of prostate cancer, to get a >15 Manchester score, would require a lot of family members to be affected

Manchester Scoring System

Cancer, age at Diagnosis	Score
♀ Breast Cancer, <30	11
♀ Breast Cancer, 30-39	8
♀ Breast Cancer, 40-49	6
♀ Breast Cancer, 50-59	4
♀ Breast Cancer, >59	2
♂ Breast Cancer, <60	13
♂ Breast Cancer, >59	10
Ovarian cancer, <60	13
Ovarian cancer, >59	10
Pancreatic cancer	1
Prostate cancer, <60	2
Prostate cancer, >59	1

A patient with breast cancer is able to get germline testing if they have:

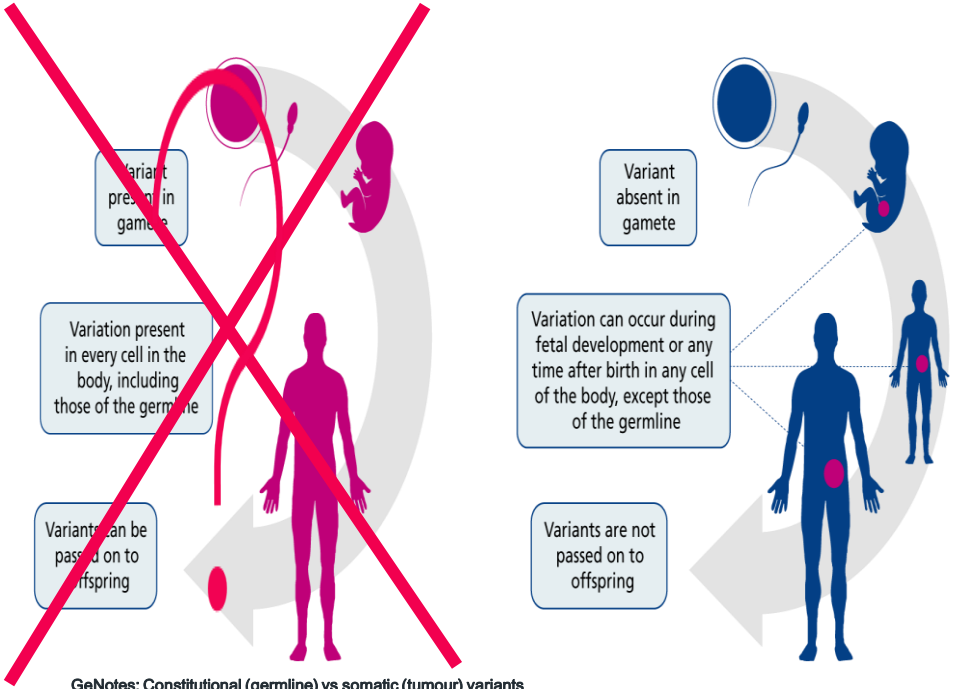
- Breast cancer ≤30
- Triple negative breast cancer diagnosed < 60
- Bilateral breast cancer both diagnosed < 50
- Breast cancer <45 and first degree relative with breast cancer <45
- Breast or ovarian cancer at any age and Jewish ancestry
- Breast cancer and Manchester Score* ≥15
- Plus other eligibility, see GeNotes for more criteria

How are we testing?

NHS East Genomics

Germline

Somatic



GeNotes: Constitutional (germline) vs somatic (tumour) variants

- Most cancer is caused by mutations that have occurred in somatic cells. These are cells that are not part of the germ cells (that produce the gametes) or gametes (egg or sperm).
- By doing somatic testing, we are testing the cancer genome.
- This enables us to detect any mutations that have been acquired during the individual's lifetime as well as those that have been inherited.
- If a mutation is detected through somatic testing, germline testing can then be offered to the patient to identify if a mutation has been acquired or inherited.
- This is important information for the patients family.

Things to consider pre testing

NHS East Genomics

The results of a test **cannot be unknown** therefore the decision to test should be taken after a full disclosure of the potential risks

A predictive or diagnostic germline test is a patient choice. Not everyone wishes to know

An understanding what interventions, if any, can be offered upon a positive result should be known.

The wait for results and the potential outcome can cause psychological worry

A negative result may not explain a strong family history therefore the preventative options may not be open to the family/patient that they wanted

The psychological impact of knowing they and their family could be at a high risk of developing cancer during their lifetime

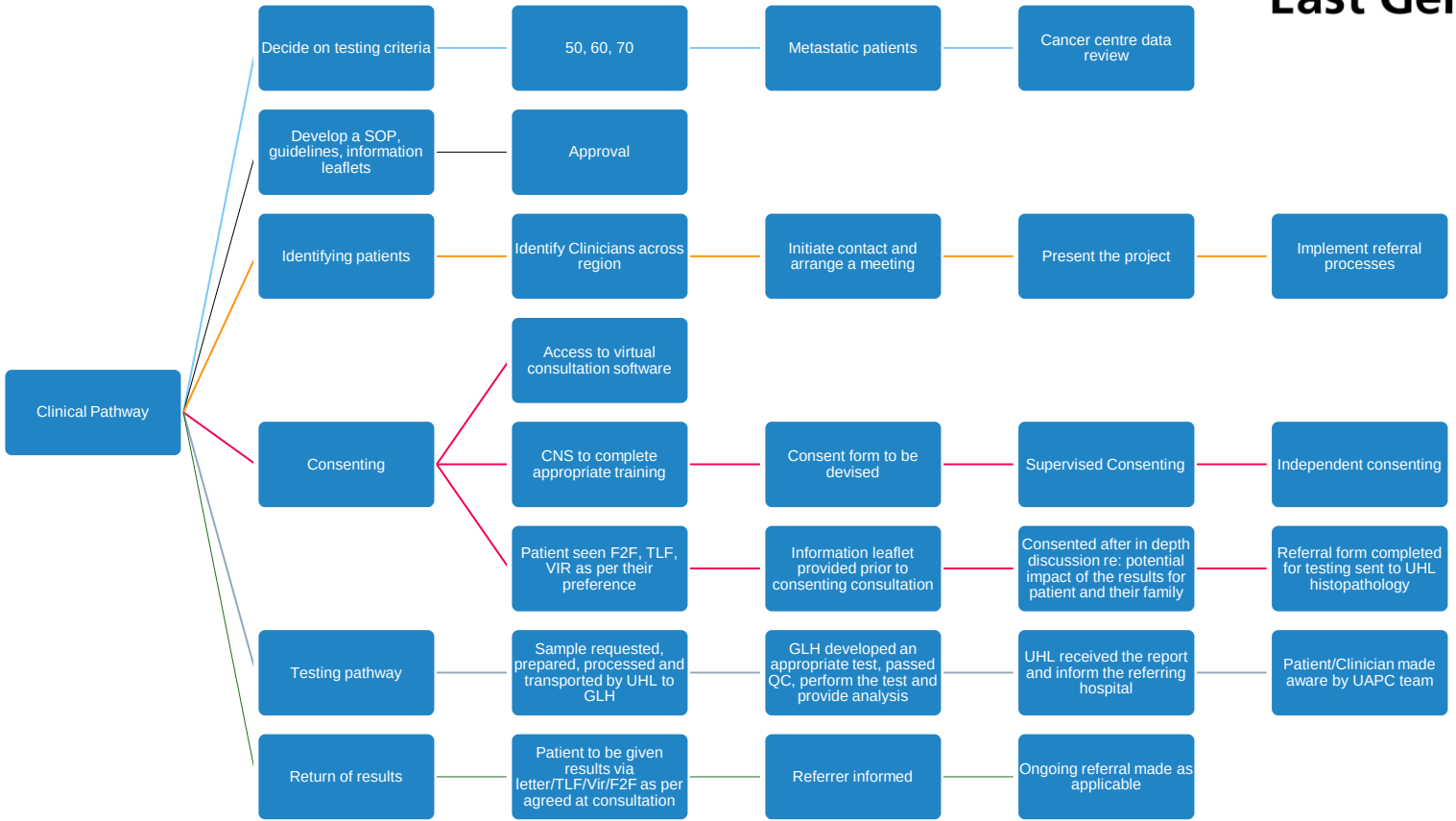
Psychological impact of a variant of unknown significance

Relationship and family dynamics can be affected

Social stigma of a diagnosis

Clinical pathway development

East Genomics



Results so far…

East Genomics

- 4 negative findings
- 2 insufficient samples (tumour at 5% and 30%)
- 1 somatic BRCA 2 mutation

- The patient with the BRCA 2 somatic mutation has now been seen in clinical genetics and has undergone germline testing.

UAPC approach to addressing the identified inequalities

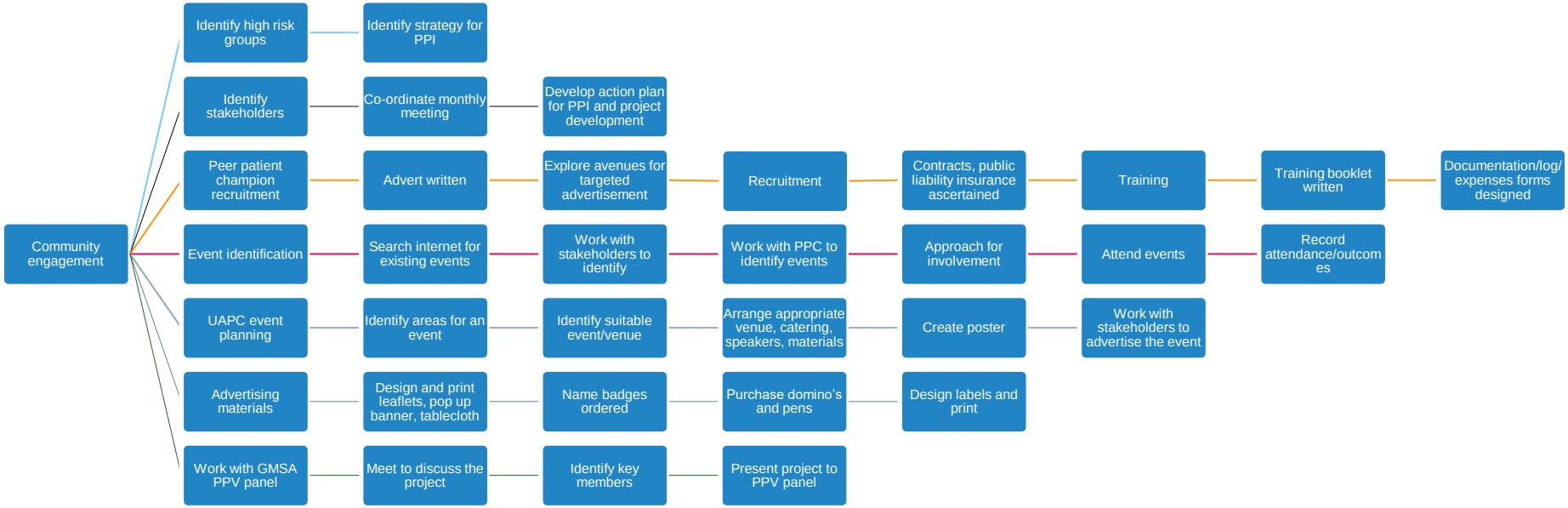
East Genomics

Clinical Pathway

Community outreach

Community engagement

East Genomics



Our stakeholders

NHS East Genomics

PROSTaid®

Is a local, registered, independent prostate & prostate cancer charity, based in Leicestershire, covering Leicestershire, Rutland and Northamptonshire. PROSTaid run support groups, offer a befriending service, fund nurse specialists, offer education and awareness raising events.

B'Me Against Cancer

Is a Nottingham charity to preserve and protect the health of people in particular but not exclusively from Black and Minority Ethnic (BME) and low-income communities living with or affected by cancer by providing and assisting in the provision of:

- culturally sensitive and appropriate practical advice, information, advocacy and support services;
- services directed to improving participation in BME cancer research and detection initiatives, and
- diversity training to improve the cultural sensitivity and service delivery of statutory bodies

The Centre for Ethnic Health Research
national centre for tackling health inequalities

The Centre for Ethnic Health Research has one clear vision: “To reduce ethnic health inequalities”. They do this by working with patients, the public, community and voluntary sectors, researchers, health and social care organisations.

UAPC resources

Pop up Banners

Tablecloth

Leaflets

Flyers

Pens

Dominos

Play domino toolkit

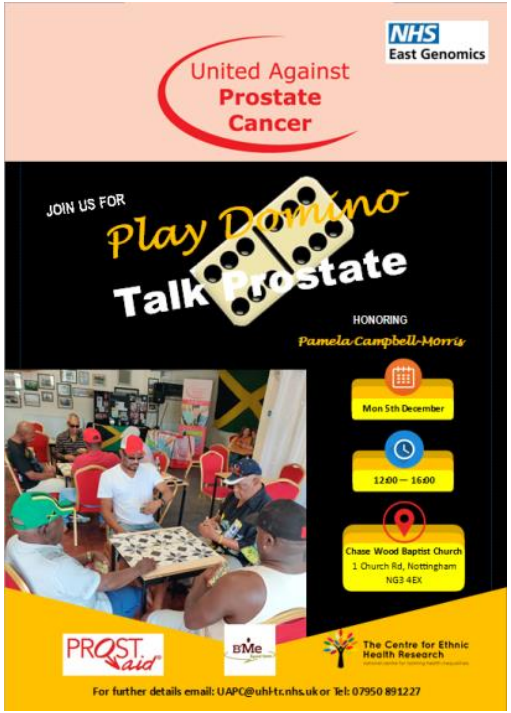
Peer Patient Champions handbook

Videos

NHS East Genomics

Play Domino Talk Prostate

NHS East Genomics

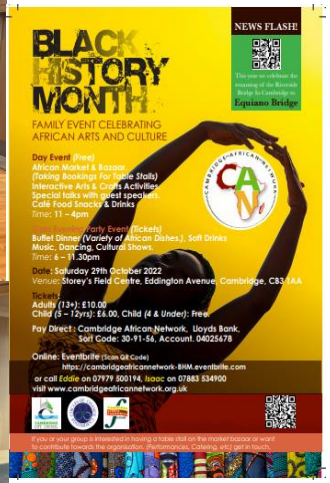


In legacy to Pamela Campbell-Morris

A relaxed, informal, culturally sensitive event engaging the community in conversations about health.

- Dominoes
- Food
- Music
- Guest speakers

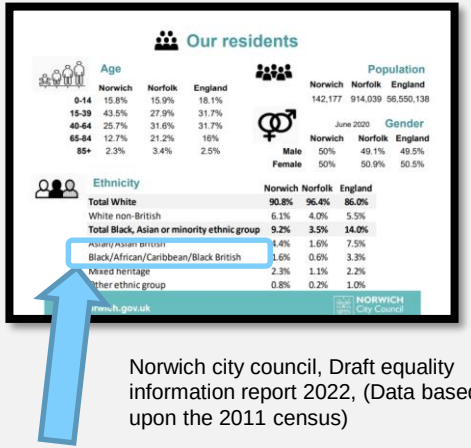
NHS
East Genomics



A few of the community engagement activities so far….

Norwich 30/09-01/10

East Genomics



Our residents

Age

	Norwich	Norfolk	England
0-14	15.8%	15.9%	18.1%
15-39	43.5%	27.9%	31.7%
40-64	25.7%	31.6%	31.7%
65-84	12.7%	21.2%	16%
85+	2.3%	3.4%	2.5%

Population

	Norwich	Norfolk	England
	142,177	914,039	56,550,138

Gender (June 2020)

	Norwich	Norfolk	England
Male	50%	49.1%	49.5%
Female	50%	50.9%	50.5%

Ethnicity

	Norwich	Norfolk	England
Total White	90.8%	96.4%	86.0%
White non-British	6.1%	4.0%	5.5%
Total Black, Asian or minority ethnic group	9.2%	3.5%	14.0%
Asian/Asian British	4.4%	1.6%	7.5%
Black/African/Caribbean/Black British	1.6%	0.6%	3.3%
Mixed heritage	2.3%	1.1%	2.2%
Other ethnic group	0.8%	0.2%	1.0%

NORWICH City Council

Norwich city council, Draft equality information report 2022, (Data based upon the 2011 census)

1.6% of the population identify as being from the African heritage community

Visitors

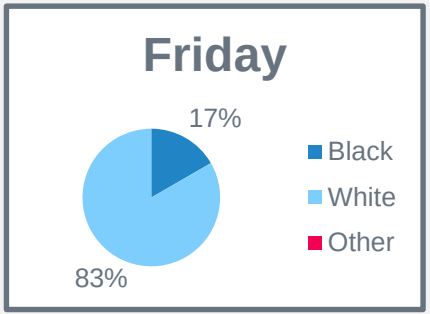
12 visitors on Friday:
7 male - 5 Female

45 on Saturday:
25 male – 20 female

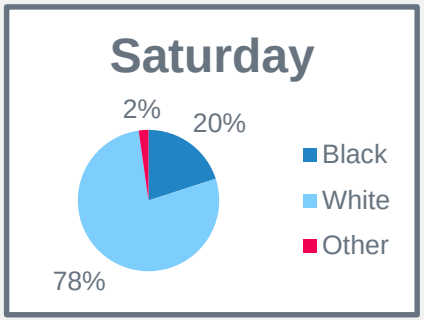
Visitors who took leaflets

Friday:	Saturday:
7 **(58%)**	23 **(51%)**

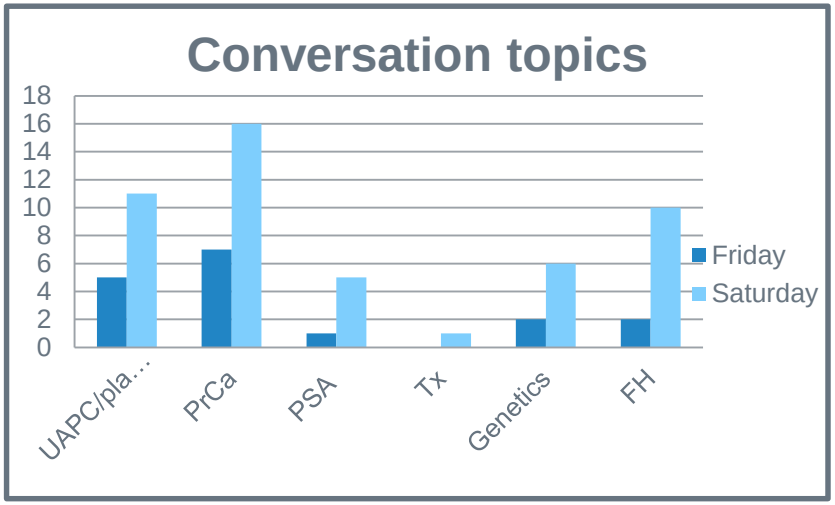
UAPC stall engagement



17% Black heritage



20% Black heritage



Signposted to:

- GP
- Healthwatch
- RMH germline testing research

"I have been and booked myself in to the GP on Monday to talk about this"

"I was told there are no symptoms of prostate cancer"

"I don't want to know"

"I've had my PSA checked every year, this year my GP stopped it. I had to push to get it done"

"I think my husband has symptoms, I'm trying to get him to go to the GP"

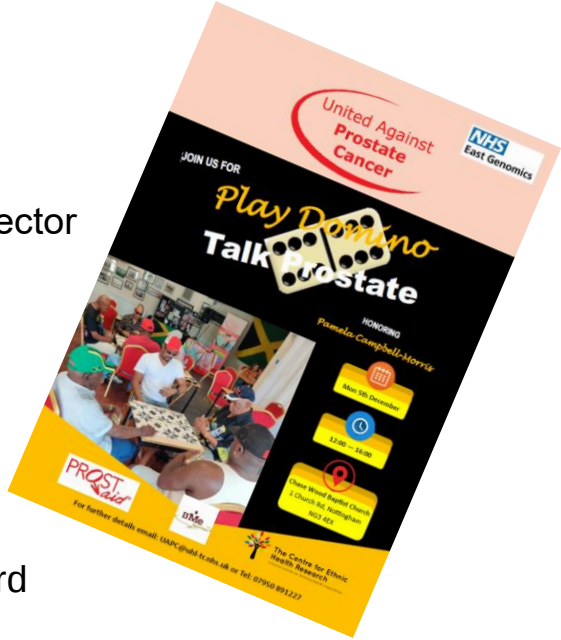
Still to come…

November:
Health inequalities meeting, Milton Keynes
Cancer risk talk, Suffolk

December:
5th Play Domino, talk prostate, Nottingham with NHS England's director for health inequalities
Cambridge African Network event

January:
Meeting with the African Caribbean organisation in Derbyshire

March:
Prostate cancer awareness event at Leicester City FC, with the Lord Mayor



Patient and public voice (PPV) panel

NHS East Genomics

- I am delighted to introduce you to Eddie Blair, who will now talk to you about his personal journey and why he is supporting UAPC today.

Thank you for listening

NHS East Genomics

Gemma Gunn
gemma.gunn@uhl-tr.nhs.uk

United Against Prostate Cancer
uapc@uhl-tr.nhs.uk

Epilepsy & Genetics

Lisa Flinton
Intellectual Disability and Epilepsy Service

Nov 2022

- Epilepsy is a disease of the brain associated with spontaneously reoccurring seizures
- Seizure; transient signs or symptoms due to abnormal excessive or synchronous neuronal activity in brain
- Epilepsy is a common brain disorder affecting 600,000 people in the UK
- Epilepsy affects people of all ages

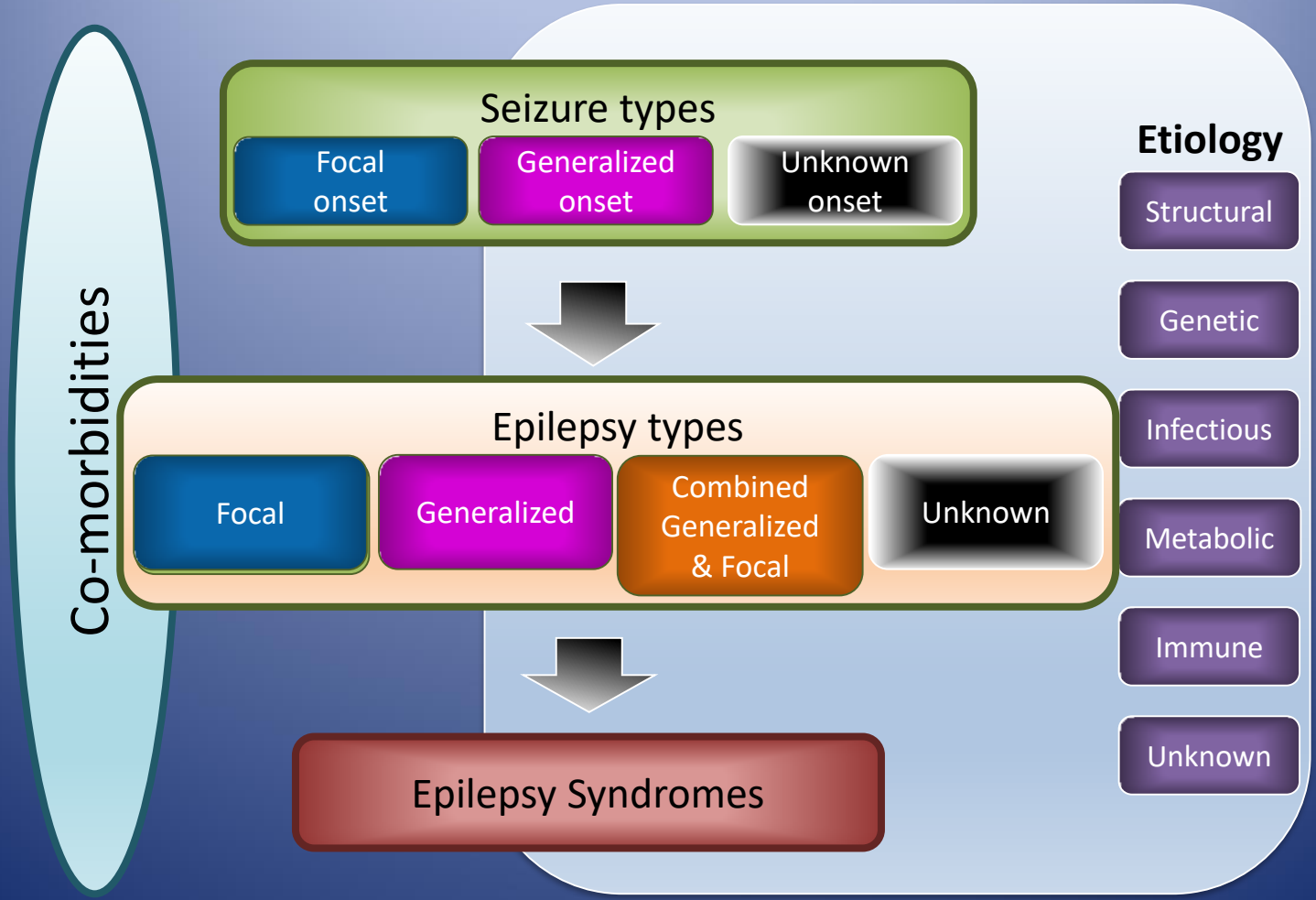
Epilepsy and intellectual disability

Commoner than in the general population

Prevalence increases with severity of disability

Often relatively severe

May be just as responsive to treatment with Specialist intervention


Co-morbidities
Seizure types
Focal onset
Generalized onset
Unknown onset
Epilepsy types
Focal
Generalized
Combined Generalized & Focal
Unknown
Epilepsy Syndromes
Etiology
Structural
Genetic
Infectious
Metabolic
Immune
Unknown

Causes of epilepsy

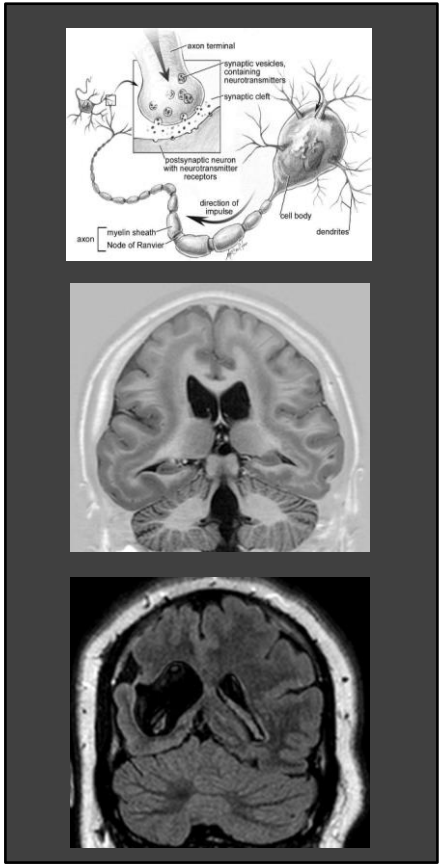
Genetic; chromosomal (Downs syndrome, XYY) or gene level (**Dravet** syndrome and channelopathies, Fragile x)

Structural; abnormality of the brain. Acquired (stroke, trauma, tumour) or genetic (tuberous sclerosis, MCD)

Metabolic; abnormalities associated with substantially increased risk of epilepsy (Glut 1, mitochondrial, lysosomal storage disorders)

Infections; in central nervous system (herpes simplex encephalitis, bacterial meningitis, HIV)

Immune; with CNS inflammation (VGKC & AMPA Receptor antibodies causing limbic encephalitis)



Genetic Disorders

- Association with learning disability
- Often dysmorphic features
- Epilepsy
- Often variable seizure types, and epilepsy usually more severe
- Rett, Angelman, Fragile X, Trisomy 21 (Downs syndrome), Ring chromosome 20

Rett UK

SUPPORT HELPLINE:
01582 798911
OTHER WAYS TO GET IN CONTACT

Rett Syndrome

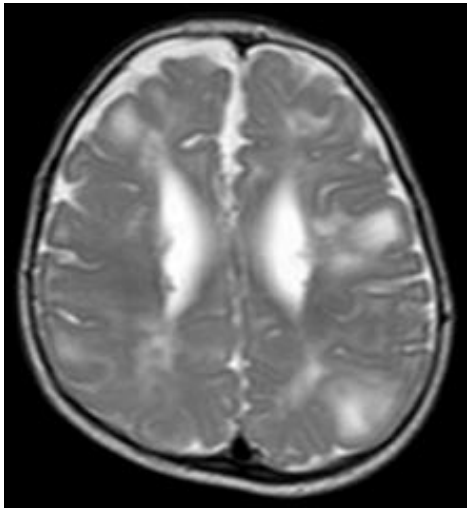
- 1 in 10,000 to 1 in 23,000 female births
- Initial normal development
- Regression from 12 to 18 months – loss of speech, purposeful hand use, withdrawal, bouts
- Epilepsy in 70 – 80%, may be focal or generalized

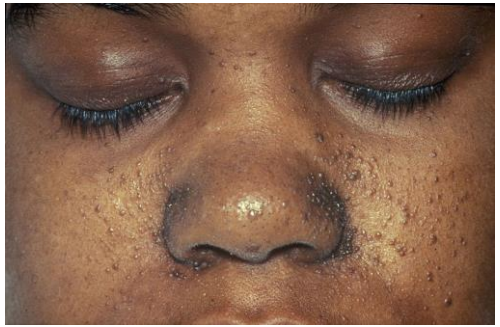
Tuberose sclerosis: Rare genetic multisystem disorder, causing tumour like lesions/hamartomas (usually benign) to develop in brain, skin and other organs.

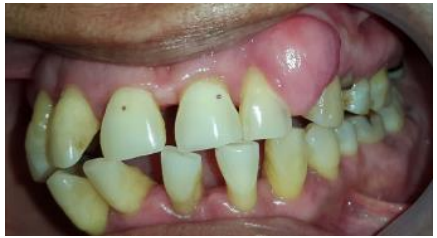
- MRI shows subependymal nodules and hamartomas (tubers). Beware SEGA can block the flow of CSF-symptoms dizziness, vomiting, reduced consciousness.
- Fibromas, angiomyolipomas in organs, skin-ash leaf and shagreen patch, LAM lungs- women avoid COCP. LD, behavioural and psychiatric probs, multifocal epilepsy
- Main cause of infantile spasms in infants. Chromosome 9 q34 encodes hamartin, Ch16 p13.3 tuberin (main cause)

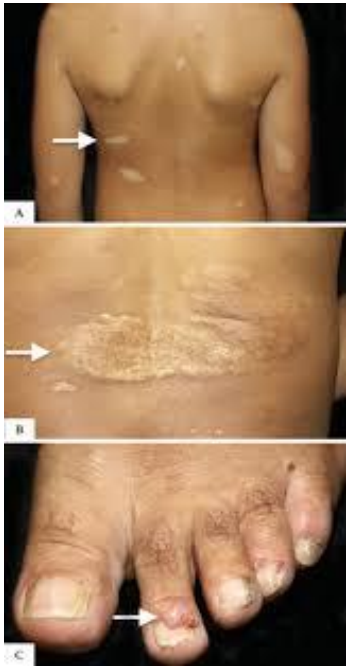
Tuberous Sclerosis

Nottinghamshire Healthcare
NHS Foundation Trust





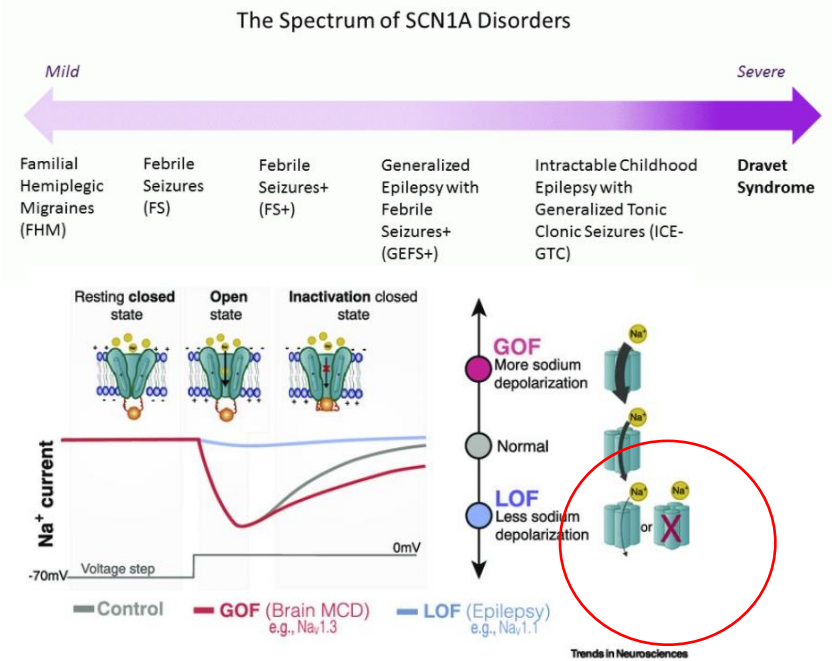




By Herbert L. Fred, MD and Hendrik A. van Dijk - http://cnx.org/content/m14895/latest/, CC BY-SA 3.0, https://commons.wikimedia.org/w/index.php?curid=11892420

Dravet Syndrome

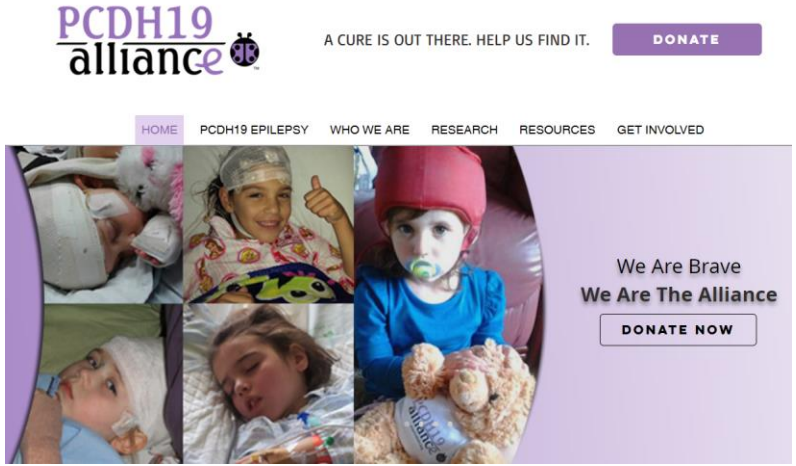
- Intractable epilepsy early life
- Seizures with fever and vaccination
- **Loss of function** of sodium channels on inhibitory neurons
- Must **avoid** common Sodium channel drugs
- Specialized drugs such as Cannabidiol and Fenfluramine
- Late complications



100,000 Genome Project

NHS
Nottinghamshire Healthcare
NHS Foundation Trust

PCDH19 (CB) normal Array.



European Journal of Paediatric Neurology 31 (2021) 61–69

Contents lists available at ScienceDirect

European Journal of Paediatric Neurology

ELSEVIER

CASK gene variant (EL) normal Array.

CASK related disorder: Epilepsy and developmental outcome

Thea Giacomini [a, b], Sara Nuovo [c], Ginevra Zanni [d], Maria Margherita Mancardi [b, *],

Take home messages

Early life epilepsy and intellectual disability often has a genetic cause

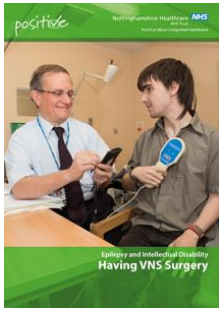
It offers the possibility of personalized medicine

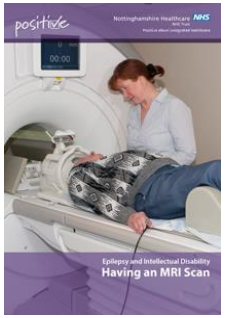
Support groups driving research

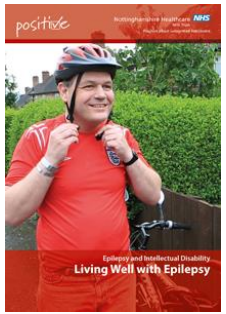
We have much to **learn**

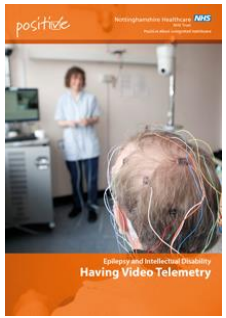
Our videos and booklets

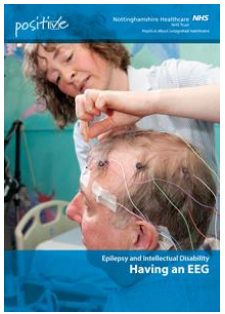
www.nottinghamshirehealthcare.nhs.uk/epilepsy











Break and networking

20 mins

Joint Nursing & Midwifery Panel Q&A

Chaired by Emma Tonkin

Final comments, thanks and close